Lisa 9

Eesti Haigekassa tervishoiuteenuste loetelu muutmise taotluse täitmisjuhis

1. Taotluse algataja	
Organisatsiooni või ühenduse nimi	Eesti Lastearstide Selts (1) Eesti Lasteneuroloogide Selts (2) Eesti Lastekirurgide Selts (3) MTÜ Eesti Seljaajusonga ja Vesipeahaigete Selts (4)
Postiaadress	Eesti Lastearstide Selts, Tartu 51014, Lunini 6 (1) Eesti Lasteneuroloogide Selts, Tartu 5014, Lunini 6 (2) Eesti Lastekirurgide Selts, Tallinn 13419, Tervise28 (3) Eesti Seljaajusonga ja Vesipeahaigete Selts, Tallinn 13419, Tervise 28. (4)
Telefoni- ja faksinumber	Tel. 6977342 (1) Tel. 7319580 (2) Tel. 6977273 (3) Tel. 6977239 (4)
E-posti aadress	info@elselts.ee (1) elns@elns.ee (2) matis@lastehaigla.ee (3) ann.paal@gmail.com (4)
Kontaktisiku ees- ja perekonnanimi ning kontaktandmed	Maila Raidmäe , maila.raidmae@lastehaigla.ee , tel. 69777127

2. Teenuse nimetus, kood ja kohaldamise tingimus(ed)	
2.1. Teenuse nimetus	**<u>Multidistsiplinaarse meeskonna (MDM) ambulatoorne vastuvõtt - uus teenusliik</u>**

	mille sisuks neuraaltoru defektiga ja sellele ligilähedaselt piiritletud patoloogiaga (perinataalperioodis omandatud hüdrotssefaalia.) haige- tele vajalik uuelaadne mitme eriala eriarsti ja/või eriala spetsialisti samaaegne meeskondlik vastuvõtt. Inimloote keskseinandi arengurikkega seotud väärarengud nagu anorektaalsed malformatsioonid, pea- ja peaaju isoleeritud arengurikked (näiteks Crouzoni tõbi jmt.), aga eelkõige kesknärvisüsteemi väärarengud Spina bifida ja HydrocephalusJa ja sellega kliiniliselt jähedane patoloogia, on multiprofiilset käsitlust va-javad seisundid – korraga on haigel koosesinevad, erinevaid organsüsteeme haaravad patoloogiad ja samaaegselt vajadus nendel kõigil erialase spetsiaalravi järele. Haige „ei ole ühegi eriala alla liigitatav" patsient, vaid uuelaadset interdistsiplinaarset käsitlemist vajav patsient. Lasteneurokirurg ravib last oma dgn-i põhiselt, silmaarst oma erialapõhiselt, ortopeed oma haigusprobleemide osas jne. vastavalt iga diagnoos vajab oma spetsialisti, oma vajalikku ja võimalikku erialaliselt heakskiidetud raviplaani kuid käsitlus peab olema terviklik ja avalduma kompleksses kõiki vajalikke erialasid kaasavas raviplaanis. /Vt. kirjandus 10. „Guidelines for Spina Bifida Health Care Services Throughout the Lifespan"./. Eesmärk on kõigi ravimiste juures haige kaasasündinud patoloogiast tulenevat malfunktsiooni ja kannatusi minimeerida ühisravimise tulemusena niivõrd, et igal haigel personaalselt oleks perspektiiv inimväärseks olemiseks, eakohaseks toimetulekuks ja sotsialiseerituseks. Kaasasündinud kesknärvisüsteemi väärarenguga kunagisest ravimatuks loetud patoloogiaga haigest on saanud **multidistsiplinaarset ravi vajav interdistsiplinaarne uuelaadne haige**, kes vajab **oma väärarengu(te) tõttu elukestvat ravi-, dispanseerse järelevalve ja jälgimise kindlustatust multidistsiplinaarse meeskonna poolt**: haige terviklik käsitlemine - **mitme eriala eriarsti ja/või eriala spetsialisti samaaegne oma liini pidi haige jätkupidev ravi, Sellise interdistsiplinaarse haige raviga seotud multidistsiplinaarse meeskonnaliikmete ühisvastuvõtt toimub haige jaoks kindla regulaarsusega haige tervisele etapphinnangu andmiseks senise ravitulemuse kohta ja edasise raviplaani ning eesmärgilisuse suhtes.** Interdistsiplinaarse haige parimad ravitulemused on, kui multidistsiplinaarne meeskond on koosseisuliselt ajateljel püsiv, kes on teadlik konkreetse (harvik)patoloogiaga haige üld- ja üksikprobleemidest, väärarengu tõsiduse ja korrigeeriva ravi nüanssidega, probleemse haigusseisu tulemuslikkusest ajateljel dünaamikas, samas ka meditsiinilises ja sotsiaalses plaanis kõik koos tervikpildina haigest. Jätkupidevalt ühed ja samad inimesed oskavad arengudünaamikat paremini hinnata, kui pidevalt vahetuv ravijate võrgustik või haige

	interdistsiplinaarsetest probleemidest mitteteadja „kõrvalseisja spetsialist / arst“. /Vt. kirjandus 20. „ Interdisziplinäre Behandlung bei Spina Bifida notwendige Versorgungsstrukturen**“,** Vt. kirjandus 1**.** *AN INTERDISCIPLINARY APPROACH, Vt. kirjandus 2. „The Turning Points conference“ 02.02. – 04.02.2017. Lieven Bauwens`i –IF SpBH peasekretär - ettekanne 03.02.2017./* Harvikpatoloogiat on vähe ning erispetsiifiline spetsiali- seerumine parimate ravitulemuste saavutamiseks koondumine referentskeskusesse on ajamärk. /*Vt. kirjandus 2. „The Turning Points conference“ 02.02. – 04.02.2017. Lieven Bauwens`i – IF SpBH peasekretär - ettekanne 03.02.2017., 21.* “ The Spina Bifida network in France”/ Läbi ajastatud regulaarselt korraldatud MDM vastuvõtu, vähemalt 3 või vajadusel enama eriarsti või spetsialisti koos meeskonnana vastuvõtt aitab tervikpilti saada haigest. Kui üks haige iseäradest on puudulikult kompenseeritud, siis tingib see ka mingi teise osa „haigestumise“ ja kokkuvõttes haige tervisseisu halvenemise või mittekompenseerituse elukvaliteedi osas.
2.2. Teenuse kood tervishoiuteenuste loetelus (*edaspidi* loetelu) olemasoleva teenuse korral	Vähemalt 3 või enama eriarsti JA spetsialisti samaaegne vastuvõtt meeskonnana (MDM = multidistsiplinaarse meeskonna vastuvõtt) DRG põhimõttel on teenus kompleksteenus, mis sisaldab endas EHK teenuskirjas olevaid teenuseid /3002,3035, 3007,3014, 7035,7050/ vastava koefitsendiga
2.3. Kohaldamise tingimus(ed)	Kõrgema etapi ravikeskus, kus on kõik olemasolevad tingimused ja erialaarstid /-spetsialistid ning võimekus korraldada kaasasündinud kesknärvisüsteemi ja sellele ligilähedase harvikpatoloogiaga haigete elukestev tänapäevanõuetele ja tasemele vastav interdistsiplinaarne ravi ning dispanseerne multidistsiplinaarne järelevalve. Kuna üldiselt on haigeid vähe ja iga haige on seejuures omakorda väga individuaalsete probleemidega vajades individuaalset sobitatud ravi ja käsitlust, siis parimad ravitulemused ongi nende haigete juures MDM hoole all kasvamine ja dispanseerne järelvalve, mis tagab ravitulemuslikkuse kompleksel ja samaaegselt arengudünaamika terviklikuse osas ülevaate koos igakülgselt erispetsiifilste osadega. Selline meeskond saab efektiivne olla kõrgema etapi raviasutuse juures, kus on võimalused kõikideks erialasteks uuringuteks ja MDM liini pidi interdistsiplinaarseks koostööks / Vt. kirjandus 10. Guidelines for Spina Bifida Health Care Services/.

2.4. Ettepaneku eesmärk	**Multidistsiplinaarse meeskonna (MDM) vastuvõtt** lisamine EHK poolt tasustavate teenuste nimekirja – **uus teenus**. Kaasasündinud kesknärvisüsteemi väärarenguga - Spina bifida, Hydrocephalus jt krooniliselt haigete elukestva jälgimise programmi „EKJP“ sisseviimiseks Eestis on väga oluline uue teenuse lisamine tervishoiuteenuste loetellu[1] ☐ Loetelus olemasoleva teenuse piirhinna muutmine[2] ☐ Teenuse kohaldamise tingimuste muutmine[3] ☐ Teenuse nimetuse muutmine[4] ☐ Teenuse kustutamine loetelust[5] ☐ Teenuse omaosaluse määra muutmine[6] **Ettepaneku eesmärk on tagada kaasasündinud kesknärvisüsteemi väärarenguga haigete elukesteva sünnist surmani elulemuse ning elukvaliteedi toetamine läbi MDM vastuvõttude** Olemasolevat kaasasündinud kesknärvisüsteemi arenguhäiret olematuks muuta pole võimalik, kuid järjepideva etappravi ning tervise arengudünaamika hindamise ja vajadusjärgse korrektiivraviga saab 1. Puudelisust vähendada (nt. hydrocepalus kontrolli alla) 2.Puudelisust kompenseerida (nt. MMC laste lihastoonuse düsbalanss kompenseerida ortoosidega) 3. Puudelisuse süvenemist mõnel juhul takistada (nt. tehtered cord`i e. seljaaju liitelise sündroomi ravi õigel ajal, liigeskontraktuuride vältimine)). Kui puudega laps sünnib, siis esmavisiidil haigevoodi juures kohtub haige ja ta pere kõigi nende spetsialistidega, kes hakkavad teda ravima ning kelle tähelepnau ja jälgimist haige oma tulevases elus edaspidi vajab. Ühiselt seatakse patsiendi jaoks eesmärgipõhine arengu-, tegevus-ja raviplaan esmaseks

[1] *Täidetakse kõik taotluse väljad või tuuakse selgitus iga mittetäidetava välja kohta, miks seda ei ole võimalik/vajalik täita.*

[2] *Kui teenuse piirhinna muutmise tingib uue meditsiiniseadme, ravimi vm lisamine teenuse kirjeldusse, täidetakse taotluses uue ressursi kohta kõik väljad või tuuakse selgitus iga vastava mittetäidetava välja juurde, miks seda ei ole võimalik/vajalik täita. Kui piirhinna muutmise tingib ressursi maksumuse muutus, täidetakse vähemalt väljad 1- 2 ning 8.1.*

[3] *Täidetakse kõik allpool esitatud väljad või tuuakse selgitus iga vastava mittetäidetava välja kohta, miks seda ei ole võimalik/vajalik täita.*

[4] *Täidetakse punktid 1-2 ning esitatakse vaba tekstina põhjendus teenuse nimetuse muutmise vajalikkuse kohta.*

[5] *Täidetakse taotluse punktid 1-2 ning esitatakse vaba tekstina põhjendus teenuse kustutamise kohta.*

[6] *Täidetakse punktid 1-2, 4.6, 9 ning esitatakse vaba tekstina põhjendus omaosaluse muutmise kohta.*

	ja samas ka pikemaks perspektiivis. Seejuures meditsiinilises erialases plaanis iga spetsialist hindab oma osa eraldi ning seab raviplaani oma erialalise vajaduse järgi kooskõlas ühtse plaaniga. Interdistsiplinaarne haigusjuhtum eeldab: I meditsiinilise poole jaoks tegutsemisjuhiseid haige ravi ning jälgimise jaoks II haigete registrit, mis võimaldaks hinnata ravi tulemuslikkust, ravi- ja profülaktikameedete sisu, mahtu, kulukut, vajadust ning planeerida sotsiaalse süsteemi kaasatuse määra abivahendite ning hoolekande korraldamisel III spetsialiseerunud multidistsiplinaarset meeskonda ja referentskeskust kõrgema etapi raviasutuse juures, kuhu harvikhaiged saab koondada, vajalikul tasemele uuringuid ja spetsiifilist eriravi korraldada ning komplekssselt ambulatoorset dispanseeringut Diagnoos **„** Spina Bifida“ , RHK nominatsioonis KAASAÜNDINUD VÄÄRARENDID, DEFORMATSIOONID JA KROMOSOOMIANOMAALIAD koodi all Q00-Q99, tähendab inimloote esmase neuraaltoru arengurikkega seotud mitut elundkonda haaravat kaasasündinud anomaaliate kompleksi, milline tingib elukestva invaliiduse suuremal või vähemal määral ja millest tervistumist ei ole. / Vt. kirjandus 5. „ SKA ekspertarstidele loeng“ dropbox aadressil slaidid 2-7 NTD patoloogia olemus /. Spina bifidat kui harvikhaiguste rühma kõige enam esinevat patoloogiat loeti aastakümnete vältel ravimatuks just uriini ja soolesisu pidamatuse, mitmet laadi neuroloogiliste ärajäämanähtude, kognitiivset laadi iseärasuste ning sageli lülisamba ja jäsemete väärmoonduste tõttu, millised enamasti tulevad esile samal ajal korraga. Selline seisund on keeruline seetõttu, et kõik haigusnähud vajavad ravimist s.t haige „ei ole kvalifitseeritav“ ühegi eriarsti ega ka üldarsti valdkonda otseselt. Samas üks diagnoos tingib teist ja nende lahenduseks tuleks ehk hoopis kolmandat ravida. Haiget ja tema seisundit tuleb hinnata ja aidata komplekssselt terviku põhimõttel, kuid iga diagnoos vajab samal ajal eriarsti erispetsiifilisi teadmisi ja nii saab haige korraga mitme eriarsti eritähelepanu keskmes olema. /Vt.kirjandus 5.„ SKA ekspertarstidele loeng“ dropbox aadressil slaid 5, 6, 7, 8,14 „ Jäädavalt on muutunud harvikhaiguste rühma liigituva Spina Bifida patoloogia, kui väga paljuprobleemse ja veel alles 90-e aastate alguses seetõttu just, ravimatuks hinnatud seisundisse suhtumine. Kaasaegsete ravivõtete ja abivahendite abil saab kesknärvisüsteemi arenguhäirega seotud haigusseisundeid edukalt ravida. Tänapäeval suudab üle 75% haigetest elada täisealiseks /Vt.kirjandus 1. „Journal of Pediatric Rehabilitation Medicine lk.1/. Reegel on see, et lapsed saadetakse kohe sündimise järel sünnitusmajast spetsialiseeritud lastekirurgia keskusesse ning ravi algab lapse sünnimomendist / Vt. kirjandus 6. ROUNDTABLE .. „Spina bifida and Hydrocephalus patient to day in Estonia“ / Seda toetab intrauteriinne diagnostika, arenemas on erispetsiifiline lootekirurgia. Kui varem loeti spina bifida seisund loote

	aborteerimise näidustseks, siis alates 2002.a. WHO direktiivi /For more information about EU rare diseases policy see http://ec.europa.eu/health/rare_diseases/policy/index_en.htm/ alusel ei kuulu Spina bifida patoloogia enam aborteerimist lubavate loote seisundite hulka, sest anomaalia on multiprofiilse õigeaegse kompleksravi abil ravitav. /Vt. kirjandus 9. Carla Verpoorten`i ettekanne/. Lapsed vajavad edasiste raviplaanide seadmiseks kohe sünnijärgselt MULTIDISTSIPLINAARSE MEESKONNA kokkusaamist haigevoodi kõrval, kus pannakse raviplaan ja järjestus paika nn. statsionaaris ex consilio esmase MDM vastuvõtuna ja edaspidi jätkupidevat juba ambulatoorses korras MDM vastuvõtuna regulaarselt haiguskulu jälgimiseks nende samade spetsialistide juures, kes on haige ja ta probleemidega tuttavad lapse sünnist saati ning antud multiprofiilset käsitlust vajava haige ravi iseärsusi tundvate erialaspetsialistidena ka oma erialaselt erispetsiifilise suunitlusega vajaliku raviplaani ja -meetmed lapsele seadnud. Kuna üldiselt on haigeid vähe ja iga haige on seejuures omakorda väga individuaalsete probleemidega vajades väga individuaalset sobitatud ravi ja käsitlust, siis parimad ravitulemused ongi nende haigete juures kasvamine MDM hoole all, mis tagab ravitulemuslikkuse arengudünaamika osas ülevaate, erispetsiifilse igakülgse ravi suunamise ja kontrolli Järjepideva igakülgse multidistsiplinaarse toetava ravi ning jälgimise abil on kesknärvisüsteemi raske väärarenguga haige võimalik arengupotentsiaal parimal moel toetatud ja esile toodud eesmärgil võimalikult iseseisvalt ja eakohaselt oma erivajadustest hoolimata elus ükskord hakkama saada/ Vt. kirjandus 10. Guidelines for Spina Bifida Health Care Services/.
2.5. Peatükk loetelus, kus teenus peaks sisalduma	Üldarstiabi X Ambulatoorne eriarstiabi ☐ Meditsiiniseadmed ja ravimid ☐ Statsionaarne eriarstiabi ☐ Uuringud ja protseduurid Operatsioonid ☐ Laboriuuringud, lahangud ja kudede transplantaadid ☐ Veretooted ja protseduurid veretoodetega ☐ Hambaravi ? ☐ Kompleksteenused ☐ Ei oska määrata/ Muu (selgitada)

3. Tõenduspõhisus ja näidustused

3.1. teenuse osutamise meditsiinilised näidustused ja vajadus teenuse osutamiseks

/ vt. Kirjandus **3.** © *1997 Rapid Science Publishers Quality of Life Research*, Vol **6**, 1997. pp. 123–132

Development of a health-related quality of life instrument for use in children with spina bifida ***/***

Spina bifida is the most common disabling congenital anomaly . However, while prevention of disabling malformations is an important goal, individuals with spina bifida often experience considerable medical and psychosocial morbidity. Spina bifida has a significant impact on the health of individuals, as is broadly defined by the concept of health-related quality of life .

/vt. Kirjandus lisaks veel 5, 10, 11- 4, 12, 15-5, 22 alt /

Kreeka keelse nimetuse ``**m**yelo**m**eningo**c**ele`` või ``meningomyelocele``(lühend **MMC)** *tõlkes myelon – seljaaju, meningo – kestad, cele – kott, kokkuvõttes seljaaju ja ta kestade songakotilaadne sopistus* on kaasasündinud esmasaju arenguhäire kompleks, mis põhjustab erinevas raskusastmes ja tasemel haigel neuroloogilisi ärajäämanähte ja arenguhäiret mitme elundkonna tasemel. Tavaliselt kaasub selja- või peaaju arenguhäirele ka aju ümbritseva luulise katte lokaalne anomaalia – väärareng. Kõige tavalisem on spina bifida e. mittesulgunud lülikaared lumbosakraalses osas, ka EESTIS. Samas Inglismaal esineb regionaalselt kõrgemad torako-tservikaalsed ajusongad ja lülisamba lülikaartelõhestumine, meie naabritel soomlaste seas on väga madalaid ja anorektaalseid malformatsioone oluliselt enam kui Eestis. Spina bifida kui esmasneuraaltoru defekt arenguanomaalia kompleksi klassifitseeritakse harvikpatoloogia gruppi, aga olles selles kõige sagedasem ja kaasaegse meditsiini võimaluste juures paremini ravitav seisund. .Neuraaltoru defekti esinemissagedus on kirjanduse järgi on erinevates riikides erinev näiteks: 4 - 4,6 juhtu 10000 elussünni kohta USA-s ; 0,9 : 10000 Kanadas; 0,7 : 10000 Prantsusmaal, 7,7 : 10000 Ühinenud Araabia Emiraatides ,11,7 : 10000 Lõuna – Ameerikas. Viimase 30 aasta jooksul on riikides, kus tegeletakse vitamiin B9 e. foolhappe profülaktikaga riiklikul tasemel vähenenud üle 70%. /vt. Kirjanduses 23. alt lisaks / Spina bifida on alati kompleksväärastustes mitmes elundsüsteemis, vahel väga kergel moel, aga enamasti raskel või väga raskel kujul. MMC haige peamine kahjustus, mis teda nn. ühiskonnast varem välja tõukas oli uriinipidamatus ja käimisvõimetus. Lisaks silmaga nähtavale jalgade deformatsioonile kaasub ~ 80%-l hydrocephalus ja üldiselt kolm aastat hilistumist kognitiivses võimekuses ja seda kuni 40 aastani. Meditsiinilist (k.a. kirurgilist) spetsiifilist eriravi ja sotsiaalse rehabilitatsioonialast sekkumist vajavad haiged alates sünnist kuni elu lõpuni. Varasemalt piirdus eluiga lapseeaga ja seepärast tundus, et antud patoloogia pole probleemne. Nüüd on aga tänud just meditsiini edusammudele eluõiguse saanud interdistsiplinaarset eriravi vajav uuelaadne haigete kontingent, kellel on tavahaigest erinevad tervisprobleemid, madaldunud võimekus enesega iseseisvalt hakkama suuremal või vähemväljendunud määral ning abivahendite ja kõrvalabi vajadus.

3.2. teenuse tõenduspõhisus avaldatud kliiniliste uuringute alusel taotletud näidustuste lõikes;

	Näide							
´ *1.*	*Editor-in-Chief.: J.A.Neufeld Journal of Pediatric Rehabilitation Medicine:*	*A*	*The Spina Bifida Association and USA National Spina Bifida programm at the Centers for Disease Con-*	*The care of children of SpB has moved beyond initial*	*To day is Focusing on those with SpB becoming fully participa-*	*By the 1970s concerns about the disabilities of SpBH children*	*Spina Bifida, Myelo-meningocele, was recog nized by the ancients. Few survived.*	*Spina Bifida Association website (sbaa.org)… This association has suppor-*

	AN INTER-DICIPLI-NARY AP-PROACH Special issue: Spina Bifida, Quest editors: Tim Brei and Sue Mukherjee Part 1 Volume1, November 4, 2008		*trol (CDC) are partnering to support quality to support quality improvments in care and increased research to improve the lives of peole with Spina Bifida*	*surgical solutions resulting in long stable periods. The cutting edge of care now includes a comprehensive team approach and a stronger emphasis on (re)habilitation.*	*ting and contributing adults in society. The single most important contribution in regards to care of he child, infant through adolescent, has been the multidisciplinary team. Pediatric specialists, pediatricians, surgeons, nurses, therapists, social workers, are all in the same place at the same time at regular intervals with good data, and are monitoring the child`s progres*	*began to mount. 70% of the that babys were selected for non-treatment.*	*The Mortality rate in the 1950-s approached 90%. To day between 70 000 and 100 000 Individuals with Spina Bifida live in the USA.*	*ted the development on the Spina Bifida Clinical Care Network with provides a foundations to enchance care along with farmework that can ultimately encourage multi-site collaborative research*

Articles by well-known cliniacians and researches on primary care, neurogenic bowel and blader, neurobehavorial ourcomes, barriers to community partipitation, adolessent, develpoment and family functioning, and the development of a bladeer protocolprogram. The cause of spina bifida is „multi-factorial". Which means that we are not sure but it appears to be some mix of nutritional and genetic factors
Vt edasi punkti 12 alt - Kasutatud kirjandus

3.3. teenuse sisaldumine punktis 3.1 esitatud näidustustel Euroopa riikides aktsepteeritud ravijuhistes;

Vt. kirjanduse alt *8. Saksamaa juhised 21. Prantsusmaa kogemus 25. Saksamaa berlini Müelodüsplaasiahaigete Keskuse lomise taotlus-põhjendus ja struktuur, kogemus 26. /19.01.2016. oli I telemiiting Eurooparriikide SpBH referentskeskuste loomise teemal, koopererumisel selleks ja võrgustiku loomine/*

Euroopa riigid alles hakkasid ravijuhist looma, aga USA on jõudnud 2 aastase suurte töögruppide najal luua üksikasjalised uued tänase meditsiinivõimalusi arvestavad ravijuhised SpBH haigete käsitlemiseks, millised peaksid maailmale kättesadaavaks ilmuma 2017.a. lõpuks.
Vt. kirjanduse alt 10. Guidelines for Spina Bifida Health Care Services Throughout the Lifespan. Council PA.
In: Merkens MJ, ed. version 3: Spina Bifida Association; 2006:70.

3.4. teenuse osutamise kogemus maailmapraktikas ja Eestis punktis 3.1 esitatud näidustuste lõikes;
Esitatakse järgmised andmed taotluse punktis 3.1. esitatud näidustuste lõikes:

a) teenuse kasutamise kogemuse kohta maailmas - mis aastast, millistes riikides teenust on osutatud ning kus riigis ja mis aastast rahastatakse antud teenust avaliku sektori poolt;
b) teenuse kasutamise kohta Eestis - mis aastast, millise tervishoiuteenuse osutaja juures on teenust osutatud, milline on teenuse maht aastas ning kas taotletavat teenust on rahastatud haigekassa poolt vm vahenditest; kui teenust on rahastatud haigekassa vahenditest, märkida, millise teenuse[7] nimetuse ja koodiga seda on raviarvel kajastatud. Kui teenust on rahastatud muudest vahenditest, selgitada millistest ning miks selline rahastamine ei jätku.

a) Vt. kirjanduse alt 10. Guidelines for Spina Bifida Health Care Services Throughout the Lifespan. Council PA.
In: Merkens MJ, ed. version 3: Spina Bifida Association; 2006:70.

b) Vt. kirjanduse alt 22. Seljaajusong ja Vesipea. Materjali koostas: lastekirurg Ann Paal Eesti Puuetega Inimeste Koja tarvis iseloomustamiseks EPIK`u allorganisatsiooni Eesti Seljaajusonga ja Vesipeahaigete Seltsi

[7] Siin ja edaspidi esitada tervishoiuteenuste loetelus sisalduvate teenuste kohta nimetus ja kood, mis on kehtestatud tervishoiuteenuste loetelus.

3.5. meditsiinilise tõenduspõhisuse võrdlus Eestis ja rahvusvaheliselt kasutatavate alternatiivsete tervishoiuteenuste, ravimite jm,;

Eriarsti teenus ja ka erispetsialistide teenused on üldjoontes olemas EHK tasustatavate teenustena ja erialaselt on ka interdistsiplinaarset harvikpatoloogiaga haiget erialaspetsialistid võimelised käsitlema, kuid see ei pruugi tulemuslik olla haige seisukohalt. Enamasti nendib „tavaspetsialist või eriarst“ multidiagnoosidega patsiendi kõrval, et kaebused on, kuid tema eriala pädevusse teiste erialade ravimine ei kuulu.Soovitatakse teise eriala spetsialisti poole pöörduda. Enamasti on igale erialale oma järjekord ning haige probleemi lahendamine jääb puudulikuks. Kõige halvem, et krooniliste haigusprbleemidega NTD patoloogia alla liigituva haige ravi ja jälgimine ei ole jätkupidev . See omakorda viib selliste keeruliste haigete juures nii ala kui ka üleravimisprobleemideni, kui ei ole haiguprobleemide dünaamika suhtes ülevaadet. Varem piirdus haigete elulemus eelkõige lapse või varase noorukieaga, sest multidistsiplinaarse haige ühtset käsitlust ei olnud veel kujunenud. Igat moodi alternatiivsed ravimeetodid üksi võetuna ei olnud tulemuslikud. Just see, et korraga oli lapsel „palju haigusi“ sai aastate eest põhjuseks, et patoloogiat loeti ravimatuks. Ühiskonnast olid haiged väljatõugatud eelkõige uriinipidamatuse ja liikumisvõimetuse pärast. Kui haige saigi kuidagi liikuma, siis muude kõrvalprobleemide tõttu laps kodusest ringist siiski kaugemale ei pääsenud. Ei olnud last näha, ei olnud ka laiemalt probleeme, mida lahendada. Selline olukord oli aastaid Eestis ja ka meist arenenuma meditsiiniga riikides, et „parem üldse mitte ravida, sest tulemust ei ole ega tule“. Tänaseks on enamasti mittemidagi tegemise seisukohtadest üle saadud nii teoorias kui praktikas. Kõiki haigeid saab kompleksravi ja õigete abivahenditega aidata. Nüüd on jõutud teise äärmusesse , et ravida me oskame ja abivahendeid on igasuguseid, kuid ravimine lihtsalt ravimisena ei pruugi soovitud loodetud edutulemust tagada ikkagi.. Tänane kaasaegne ravi ja abivahendid on muutunud nii kalliks, et kerkivad mitmet laadi eetilised küsimused, kas need igasugusele haigele on vajalikud ja sobivad. Esiplaanile on tõusnud ennetavad profülaktilised meetmed, et üleüldse sünniksid terved beebid. Kui juba on siiski olemas interdistsiplinaarne multidiagnoosidega haige, siis kordades on odavam õigeaegne igakülgne ravi, kui hilistunud sekkumine või üldse mitte sekkumine ja tüsistuste ravi. *Vt. kirjanduse alt 10. Guidelines for Spina Bifida Health Care Services Throughout the Lifespan. Council PA. In: Merkens MJ, ed. version 3: Spina Bifida Association; 2006:70. Vt. kirjanduse alt 9. Carla Verpoorten – Belgia Ghenti Ülikooli lasteneuroloog`i loeng IF SpBH seminaril 03.02.2017. Tase D alternatiivsete meedtete võimalikkuse kohta*

3.6. teenuse seos kehtiva loeteluga;

MDM vastuvõtt on lahtikirjutatav EHK teenuskirjas olevate teenuste /3002 – eriarsti esmane vastuvõtt 20,15 EUR-i, 3035 – õe iseseisev vastuvõtt 10,39 EUR-i, 3007 – kliinilise psühholoogi vastvõtt eriarsti suunamisel 22,31 EUR-i, 3014 – logopeedi vastvõtt eriarsti suunamisel 27,49 EUR-i, 7050 – füsioteraapia individuaalne tegelus 30 min. 11,15 EUR-i/ põhjal , 7053 – tegevusterapeudi teenus 30 min. 12,07 EUR-i, mis aga oleksid vastava koefitsiendiga uues 1 tunnises kompleksteenuses. Uudne

multidistsiplinaarse meeskonna vastuvõtt võiks olla ka näiteks DRG piirhinna põhimõttel lahtikirjutatud.

3.7. teenuse seos erinevate erialade ja teenuse tüüpidega;
NTD EKJP- elukestvalt krooniliselt haigete jälgimise polikliiniku meeskond:
(Eri)ARST -/neuroloog (eelistatult), kirurg või sisearst/ – ravijuht (ingl. keeles ***Case manager)***,
õde - koordinaator, arsti abiline,
eriarst id - (neuro)kirurg, ortopeed, nefroloog(uroloog), arst, keda kaastakse vastavalt vajadusele nagu psühhiaater, silmaarst, kõrva-nina-kurguarst, endokrinoloog, kardioloog põhimeeskonda täiendavad liikmed vastavalt juhtiva patoloogia vajadusele
spetsialistid - füsioterapeut, psühholoog, sotsiaaltöötaja, ortoosimeister, logopeed(eripedagoog), sotsiaaltöötaja

ARST – uuelaadne eriarst – on kogu üksuse juht ja haige jaoks meditsiinilise tervisloo suunaja-hindaja, krooniliselt haige järjepidevuse ülevaataja

Nõuded MDM VASTUVÕTU juures arstile :

- Patoloogia põhjalik tundmine ja erialane pädevus,
- Väärarengu etioloogia ning patofüsioloogia tundmine,
- Patoloogia ravipõhimõtete ja maailma meditsiiniteadmistega kursis olemine
- Teadmine optimaalsetest raviaegadest, kas operatiivne või konservatiivne sekkumine või alternatiivsed ravimeetodid oleksid juhtumipõhiselt parimad
- Teab üksikasjaliselt dispanseersel jälgimisel oleva haige kohast informatsiooni järjepidevuse osas ja interdistsiplinaarselt
- annab sellest informatsiooni perekonnale ning patsiendile endale
- Koordineerib pere jaoks kogu info saamist ja interdistsiplinaarset järelvalvet / ravi, suhtleb kolleegidega, MDM vastvõtu läbiviija ja tulemuslikkuse hindaja

- Vahendaja – koordinaator meeskonna ja perekond + haige ja haige teiste sidusgruppide – kool, lasteaed jm. - vahel
- Tunneb lapseea arenguetappe ja ravisekkumiseks optimaalset aega, on kursis sama patoloogia täiskasvanuteks sirgunud patsientide probleemidega
- Kaasasündinud kesknärvisüsteemi ja sellele sarnaste patoloogiaga haigete – krooniliste probleemidega haigete juures tuleb arvestada kõigi haigel olevate diagnoosidega ja ka nende osas kaasasündinud iseärasustega, millistest üks või teine, kolmas, neljas vajab pidevat kompensatoorset ravi või jälgimist, eriravivajaduse kujunemise või aegajalt tsüklilist eriravi
- Mitmed neuroinfektsioonid, viirushaigused nagu gripp jt, nakkushaigused võivad kulgeda NTD haigetel tavanormsest tõsisemate tagajärgedega. Krooniliselt haige algselt muutunud orgaanilise leiu dünaamikat ägeda muu haigusega seotud muutusi suudab parimal moel jälgida ja ravida NTD patoloogiat tundev ja haige põhiprobleemide järjepidevusest ülevaadet omav MDM meeskonda kuuluv arst (neuroloog). On erinevaid praktikaid juhtiva arsti erialase kuulumise suhtes. Näiteks

Eestis, Tšehhis, Soomes Oulu prk-as on siiani olnud lastekirurg, Hispaanias, Saksamaal, Belgias neuroloog, Kanadas ja USA-s terapeut

ÕDE –koordinaator, arstiabiline

Nõuded MDM VASTUVÕTU juures õele :

- Peab registrit
- Teeb registreeringuid etteantud programmi alusel
- Valmistab sisuliselt dokumendid MDM vastuvõtule kutsutud patsiendi kohta ja vastuvõtu ajal aitab, korraldab, vormistab, suunamisi ja uuringuid jne. olles otsene arsti abiline
- Õde – koordinaatori kaudu korraldatakse konkreetsed tähtajad uuringutele, visiitidele eriarstide või spetsialistide juurde teavitatakse haiget edasistest visiidiaegadest
- Aitab erakorraliste probleemide esilekerkimisel olukorrale vastavalt juhiste või tarviliku info esitamisega haiget või tema heaolu ja tervisega seotud teist isikut, abistab arsti patsiendi läbivaatuse ja protseduuride juures
- Reeglina on esimeseks kontaktisikuks kõigile EKJK – elukestva jälgimise keskuse - registris olijatele, kui patsiendil või ta lähedasel, sidusisikutel peaks olema küsimusi või probleeme millele nad ootaksid vastust või lahendust Näiteks tervisalane nõustamine, abivahendite kasutamine ja saamine jmt.
- On isikuandmekaitse eest vastutaja samaväärselt arstiga
- On kontaktisikuks spetsialistide vahel, vastuvõtule ja uuringutele saatmiste korraldaja
- On pädev iseseisvalt töötama näiteks troofiliste haavandite hooldusravi, uroteraapias põie isekateeterdamise probleemid, toitumissoovitused, nõustamiste ja kooli või lasteaia sobitamise küsimuste alal. Oskab probleemi sügavust hinnata ja vajadusel oma pädevust ületavate küsimuste korral õigesse kohta abi saamiseks pöörduda või patsienti suunata

ERIARST

Nõuded MDM VASTUVÕTU juures (eri)arstile :

- NTD kui harvikpatoloogia põhjalik tundmine ja erialane pädevus,
- Väärarengu etioloogia ning patofüsioloogia tundmine,
- NTD kui harvikpatoloogia praegusaegsete ravipõhimõtete ja maailma meditsiiniteadmistega kursis olemine
- Teadmine optimaalsetest raviaegadest, kas operatiivne või konservatiivne sekkumine või alternatiivsed ravimeetodid oleksid juhtumipõhiselt parimad oma eriala spetsiifika ja seejuures ka interdistsiplinaarse haige juures
- Teab üksikasjaliselt dispanseersel jälgimisel oleva haige kohast informatsiooni oma erialase liini pidi järjepidevalt ja

interdistsiplinaarse haige arengudünaamika ning kasvamise kohalt, annab sellest informatsiooni perekonnale ning patsiendile endale

- Omab pädevust oma eriala liinis kui MDM töö liinis olles partneriks kõigile meeskonna liikmetele nii samaaegselt kui ka järjepidevalt. Tunneb lapseea arenguetappe ja ravisekkumiste optimaalset aega oma eriala liinis ja suudab kohandada parimate lahenduste ning ajatamistena oma eriala kompetentsi NTD patoloogia – haige - multiprobleemide suhtes. On kursis oma erialselt patoloogiaga täiskasvanuteks sirgunud patsientide probleemidega kui järjepidevust järgivalt ka MDM hoolealuse arengudünaamikaga
- Kaasasündinud kesknärvisüsteemi ja sellele sarnaste patoloogiaga haigete – krooniliste probleemidega haigete juures tuleb arvestada kõigi haigel olevate diagnoosidega ja ka nendel oleva iga diagnoosi kaasasündinud iseärasustega, millistest üks või mitu vajab vahel pidevat kompensatoorset ravi, kolmas diagnoos jälgimist eriravivajaduse kujunemise seisukohalt, mõni haigusseisu kompleksdiagnoos ainult aegajalt tsüklilist eriravi
- NTD patoloogiat tundev ja haige põhiprobleemide järjepidevusest ülevaadet omav spetsiifiliste erialaste teadmistega MDM meeskonda kuuluv eriarst on partneriks kõigile interdistsiplinaarse haigega seotud osapooltele

- *<u>KIRURG</u> - on NTD interdistsiplinaarses meeskonnas juhtiv spetsialist, sest temast sõltub kõige enam haige puudelisuse korrektiivravis elukestvalt / esmane kirurgiline korrektsioon, jätkukirurgilised vahelesekkumised, lapse kasvamisega seotud kirurgilised korrektsioonid, ägedad kirurgilised patoloogiad, HC shundiga seotud probleemid, traumapuhune spetsiifiline ravi / näiteks luumurrud Spina bifida haigel lapsel jmt./, tethered cord, troofilised haavandid*

- *<u>ORTOPEED</u> – NTD laste ortopeedia probleemid on erisugused, millega peab arvestama, ortopeed on patoloogia spetsiifika ekspert ja tihedas koostöös meeskonna teiste liikmetega*

- *<u>SILMAARST</u> - paljud lapsed vajavad prille, HC shundiklapirõhu mittesobilikkus võib avalduda paispapilli tekkena jm*

- *<u>KNK ARST</u> - kuulmisvõimekus võib olla langenud*

- *<u>PSÜHHIAATER</u> – suurte keskuste analüüsiandmetel on kaasasündinud kesknärvisüsteemi patoloogiaga krooniliselt haigete laste võimekuse ja just kognitiivses plaanis võimekuse areng selektiivset laadi ning keskmiselt 3 aastase hilistumisega ning kohati sügava mahajäämuseni*

- *<u>ENDOKRINOLOOG</u> – peaaju kestseinandi ärritusnähu üheks väljenduseks on endokrinoloogilist laadi kõrvalekalded nagu liiga väike kasv, liiga kiire kasv või pubertas praecox, kilpnäärme talitushäired, mis vajavad järjepidevat seiret ja vajadusele vastavat toetusravi*

- *<u>NEUROLOOG</u> – halvatusnähud, nahatroofika ja tundehäired, epilepsia vajab väga tähelepanelikku spetsiifilist eriravi ja*

jälgimist dünaamikas elukestvalt

- *<u>SISEARST</u> – pidev ravimite võtmine võib mõjutad amao-seedetrakti, toitumisprobleemid, neurogeense soole probleemist kõhukinnisuse ja intoksikatsiooni võimalus, vererõhu ja kardiopulmonaalsed probleemid esinevad sagedamini tervetest*

- *<u>UROLOOG / NEFROLOOG</u>- neurogeense põie tõttu on ülisuur oht neerupuudulikkuse kujunemisele, kui ei ole adekvaatset uropassaaži ravi seatud või on urotrakti raske väärareng, mis esineb antud haigete grupil kõrvalpatoloogiatest tihti*

SPETSIALIST

<u>Nõuded MDM VASTUVÕTU juures spetsialistile</u> :

- NTD kui harvikpatoloogia põhjalik tundmine ja erialane pädevus,
- Väärarengu etioloogia ning patofüsioloogia tundmine,
- NTD kui harvikpatoloogia praegusaegsete ravipõhimõtete ja maailma meditsiiniteadmistega kursis olemine
- Teadmine optimaalsetest raviaegadest, sekkumiste võimalustest ja alternatiivsetest ravimeetodidest oleksid juhtumipõhiselt parimad oma eriala spetsiifika ja seejuures ka interdistsiplinaarse haige juures
- Teab üksikasjaliselt dispanseersel jälgimisel oleva haige kohast informatsiooni oma erialase liini pidi järjepidevalt ja interdistsiplinaarse haige arengudünaamika ning kasvamise kohalt, annab sellest informatsiooni perekonnale ning patsiendile endale
- Omab pädevust oma eriala liinis kui MDM töö liinis olles partneriks kõigile meeskonna liikmetele nii samaaegselt kui ka järjepidevalt. Tunneb lapseea arenguetappe ja ravisekkumiste optimaalset aega oma eriala liinis ja suudab kohandada parimate lahenduste ning ajatamistena oma eriala kompetentsi NTD patoloogiaga patsiendi multiprobleemide suhtes. On kursis oma erialselt patoloogiaga täiskasvanuteks sirgunud patsientide probleemidega järjepidevust järgivalt
- Kaasasündinud kesknärvisüsteemi ja sellele sarnaste patoloogiaga haigete – krooniliste probleemidega haigete juures tuleb arvestada kõigi haigel olevate diagnoosidega ja ka nende iga diagnoosi kaasasündinud iseärasustega, millistest üks või mitu vajab vahel pidevat kompensatoorset ravi, kolmas diagnoos jälgimist eriravivajaduse kujunemise seisukohalt, mõni haigusseisu kompleksdiagnoos ainult aegajalt tsüklilist eriravi
- NTD patoloogiat tundev ja haige põhiprobleemide järjepidevusest ülevaadet omav spetsiifiliste erialaste teadmistega MDM meeskonda kuuluv spetsialist on partneriks kõigile interdistsiplinaarse haigega seotud osapooltele

- *<u>FÜSIOTERAPEUT</u> - on tihedalt koos neuroloogi ja ortopeediga, kirurgidega, kes on võtmeisikuks haigete liikuma hakkamise ja arengu suunamisel. Füsioterapeut tegeleb haigetega vastavalt vajadusele omaette koostöös eriarstidegaja*

ortoosimeistriga, aitab korraldada liikumist ja toimetulekut hõlbustavaid abivahendeid

- *ORTOOSIMEISTER – NTD patoloogia haigete tarvis eriettevalmistuse saanud meister, kes valmistab lihasdüsbalansi korrigeerimiseks ortoose, vaatab dünaamikas nende sobilikkust, korrastab ja täiendab või teeb vajadusel uued, sobitab liikumise tarvis ja asendeid korrigeerivaid abivahendeid*

- *SOTSIAALTÖÖTAJA – on sotsiaalse sidususe ekspert ja abiline haige, administratiivsete asjaajamiste ja võimaluste leidmise jaoks. Laste puhul ka nende vanematele seaduste ja infokättetooja, noortele täiskasvanudhaigetele töötamise ning võimalikult iseseisva toimetulekuvõimaluste leidmise tugi*

- *LOGOPEED - keelelise ja kognitiivse võimekuse arendamise eesmärgil vajavad peaaegu kõik NTD patoloogiaga krooniliste multidiagnoosidega interdistsiplinaarsed haiged logopeedilist õpetust kõne arengu, lugema õppimise ja esinemisoskuse ning suhtlemise arenemise juurde*

- *PSÜHHOLOOG – kesknärvisüsteemi ja keskseinandi kaasasündinud patoloogiale kaasub suuremal või vähemal määral peaaju ja neurotraktide anormsus, mis on seotud looteea arengu iseäraga ja sellest tulenevalt on ka hilisemas elus psüühika ja kognitiivse võimekuse iseärasid antud haigete grupis arvatust suuremal hulgal tervete eakaaslastega vrd-s*

4. Teenusest saadav tulemus ja kõrvaltoimed

4.1. teenuse oodatavad tulemused ning nende võrdlus punktis 3.2 esitatud alternatiividega (ravi tulemuslikkuse lühi- ja pikaajaline prognoos:

Tänapäevane ravitaktika on, et intrauteriinselt diagnoositud SpB või HC laps sünniks keiserlõike teel võimalikult ajaliselt küpses vanuses. Lapse spetsiifiline eriravi hakkab sünnijärgsena kohe multidistsiplinaarse meeskonna poolt seatud raviplaani alusel ja sama meeskond jääb last jätkupidevalt regulaarselt jälgima ning kindla regulaarsusega kokku saama. Tähtis on , et laps areneks dünaamiliselt kõigi erialaspetsialistide poolselt jätkupidevalt hinnatud ja suunatud. Eesmärgiks on lapse kompleksravis lapse areng võimalikult eakohase normi keskmisele tasemele ja iseseisvale toimetulekule. TÄNA EI LOETA PATOLOOGIAT RAVIMATUKS. Elulemus on perspektiivis sügava pensionieani. Täisealiseks saanud NTD patsientidel on rida spetsiifilisi probleeme, milliste minimaliseerimine on ja jääb suuresti sotsiaalsüsteemi kanda. Meditsiini liinile jääb 50%, mistõttu MULTIDISTSIPLINAARSE MEESKONNA KONTROLLIV ja SUUNAV ning ABISTAV ROLL ON HAIGETELE ELUKESTVALT VAJALIK MEEDE inimväärse elukvaliteedi hoidmiseks. Haigete töövõimekus on ja jääb teatud määral madalamaks eakaaslastega vrd-s, aga enamus on sobivate toetavate struktuuride olemasolul võimelised iseseisvaks eluks ja

töötamiseks. Teatud hilistunud arengujoonega tuleb arvestada vähemalt 40 . eluaastani.
MDM on NTD patoloogia haigete grupile tugisüsteemiks nende tervisseisu stabiilsel rahuldaval tasemel toetamisel.

4.2.
Prognoos on kestvaks eluks hea vaatamata hulgipatoloogiate esinemisele. MDM põhimõttel haigete dispanseering, jälgimine ja toetavate otseste ravimeedte juurde õigeaegne suunamine ning igasuguste võimalike tüsistuste profülaktika on MDM peamine eesmärk.
Üldine prognoos täisealistele on, et vaatamata oma erivajadustele ja abivahenditele (suurte keskuste statistika USA, Orlando, 2009.a.mai, I SpB maailmakongress)
30% suudab iseseisvalt elus toime tulla
45% vajab oasliselt abi ja toetamist
20% saab hakkama ainult pideva kõrvalabiga
5% on rasked hooldatavad patsiendid

4.3. teenuse kõrvaltoimed ja tüsistused, nende võrdlus punktis 3.2 esitatud alternatiivide kõrvaltoimetega:

Taotletaval teenusel kõrvaltoimed ja tüsistused puuduvad

4.4. punktis 4.2. ja 3.5 esitatud kõrvaltoimete ja tüsistuste ravi kirjeldus (kasutatavad tervishoiuteenused ja/või ravimid (k.a ambulatoorsed ravimid));

Esitatakse loetelu teenustest ja ravimitest (loetelus sisalduvate teenuste ja ravimite puhul esitatakse nende nimetus loetelus kehtestatud kujul ning lisatakse juurde ka kood), mida on vajalik patsiendile osutada, et ravida tekkinud kõrvaltoimeid ning tüsistusi. Lisaks esitatakse nimetatud teenuste ja ravimite lõikes keskmine teenuste kasutuskordade arv ning keskmised ravimite annuste suurused ühe isiku kohta.
Näiteks:
Teenusega kaasneva tüsistuse z raviks kasutatakse:
1) ravimit q, keskmine kogus patsiendi raviks 10 ühikut,
2) protseduuri w, üks kord patsiendi kohta.

4.5. taotletava teenuse osutamiseks ja patsiendi edasiseks jälgimiseks vajalikud tervishoiuteenused ja ravimid (s.h ambulatoorsed) vm ühe isiku kohta kuni vajaduse lõppemiseni ning võrdlus punktis 3.5 nimetatud alternatiividega kaasnevate teenustega;

Multidistsiplinaarne meeskonna vastuvõtt – teenusena oleks meditsiinisüsteemis ambulatoorses praktikas eriarsti/de ja erispetsialistide kompleksteenus kõrgema etapi raviasutuse juurde koondatud harvikhaigete /NTD patoloogia alla liigituvate diagnoosiga haigete jaoks.

Teenuse aluseks on baasmudel, kus ühe teenustunni osutamisse on integreeritud EHK rahastatavad teenusted:
minimaalselt 2-3 või vajadusel enamat eriarsti, 2 või enamast erispetsialististi ja arvestatud eraldi asjaajaja-sekretär
Teenust osutava personaliga on seotud põhitöötajate töötasud, töövahendid, töökoht, koolituskulude ja supervisiooni komponent, büroo- ja sidekulud. Patsiendi teenindamisega on seonduv inventar, seadmed. Sellele lisanduvad lisakulud ja asutuse üldkulud.

4.6. teenuse võimalik väär-, ala- ja liigkasutamine; teenuse optimaalse ja ohutu kasutamise tagamiseks teenusele kohaldamise tingimuste seadmise vajalikkus;

Teenuse väärkasutuse ennetamiseks on vajalik piirata teenuse osutajate hulka piirkondlike kõrgema etapi haiglatega : Tallinna Lastehaigla , edaspidi teenuse laienedes täiskasvanutele lisandub Põhja-Eesti Regionaalhaigla, Tartu Ülikooli Kliinikum.
Teenuse sagedus määratakse juhendiga vastavalt vanusegrupile

4.7. patsiendi isikupära võimalik mõju ravi tulemustele;

Lapseealine sõltub vanematest ja hooldajatest. Täiskasvanute juures on vajalik perearsti, tugiisikute jt. informeeritus ning abi MDM vastuvõttudel patsiendi regulaarse käimise / toomiste juurde, kui tal enesel puudub selleks võimekus, MDM otsuste ja soovituste järgmise tarvis. Üldplaanis on kesknärvisüsteemi kaasasündinud väärareng harvikpatoloogia ja alati multiprobleemne ning elukestev. *Vt. kirjanduse alt 10. Guidelines for Spina Bifida Health Care Services Throughout the Lifespan. Council PA. In: Merkens MJ, ed. version 3: Spina Bifida Association; 2006:70.* **Tase A** *Vt. kirjanduse alt 11. Second World Congress on Spina Bifida Research and Care 2012 . USA, Las Vegas Urology Abstracts. " Medical and Surgical Management of Adult Patients with Spina Bifida"* Author: Jessica Casey, MS, MD, Northwestern Memorial Hospital USA, Department of Urology
....Medical management of spina bifida patients is commonly complicated by their multiple medical co-morbidities....

5. **Vajadus**

5.1. Eestis teenust vajavate patsientide arvu hinnang (ühe aasta kohta 4 aasta lõikes), kellele on reaalselt võimalik teenust osutada taotletud näidustuste lõikes:

Esitatakse punktis 3.1 esitatud näidustuste lõikes patsientide arv järgneva nelja aasta kohta eraldi, kellele on reaalselt võimalik teenust osutada (arvestades teenust vajavate patsientide arvu, teenuse osutajate arvu, vajalikku infrastruktuuri jm). Esitatakse

andmed, mille põhjal on teenust vajavate patsientide arvu hinnatud ning selgitused patsientide arvu muutumise kohta aastate lõikes. Patsientide arv esitatakse alljärgnevas tabelis:

Teenuse näidustus	**Patsientide arv aastal *t****	**Patsientide arv aastal *t+1***	**Patsientide arv aastal *t+2***	**Patsientide arv aastal *t+3***
1	**2**	**3**	**4**	**5**
Esitatakse teenuse osutamise näidustus kehtiva haiguste klassfikatsiooni alusel	*Märgitakse teenust vajavate isikute arv veerus 1 esitatud näidustusel aastal t*	*Märgitakse teenust vajavate isikute arv veerus 1 esitatud näidustusel aastal t+1*	*Märgitakse teenust vajavate isikute arv veerus 1 esitatud näidustusel aastal t+2*	*Märgitakse teenust vajavate isikute arv veerus 1 esitatud näidustusel aastal t+3*

**t* – taotluse menetlemise aastale järgnev aasta;

1a	5		1
2a	0		5
3a	2		1
4a	6		2
5a	3		7
6a	5		6
7a	3		5
8a	3		3
9a	4		3
10a	3		5
11a	5		4
12a	7		5

13a	5		6
14a	8		6
15a	8		9
16a	13		9
17a	8		5
18a	6		5
	94		**87**

SpinaBifida **Hydrocephalus**

Q05 **Q03**

~5 uut juhtu aastas ~ 5uut juhtu aastas

1998 - 2016 TLH andmete analüüsi alusel

Teenuse kasutamise sagedus vastavalt eagruppidele

1-12kuud	**1-5a**	**6-18a**	
VS	2 x aastas	1 x aastas	
3k			
12k			
5last	4 x 5 last = 25 last	12 x 5 last= 60 last	90 last
5x3= 15 korda	25 x 2 = 50 korda	60 korda	

Aastas 125 korda meeskonna kokkusaamisi
a` 60 min = 125 tundi MDM visiiti. Lisama peaks , et kõikumise juures ca 5% ulatses s.o. 5-6 haiget siia-sinna tuleb arvestada ~ 130 MDM visiidga laps kontingendile Kui

praeguse sündimuse juures lisandub aastas 6 – 7 uut temaatilist kroonilist haiget, siis järgmisel aastal on vajalik nende lisamine praeguse ~130 visiitide arvule.
Täiskasvanuks saanute patsientide tarvis tuleks planeerida samuti võimalus ~170 MDM visiidi jaoks sama hinnakirja alusel. Seega aastas kataks praeguse NTD patoloogiaga haigete arvukuse juures 300 MDM visiiti aastas nende vajadused

5.2. teenuse mahu prognoos ühe aasta kohta 4 järgneva aasta kohta näidustuste lõikes:
Esitatakse andmed prognoositava teenuse mahu kohta näidustuse lõikes järgneva nelja aasta kohta iga aasta kohta eraldi, arvestades teenust vajavate isikute arvu ning keskmist teenuse kasutuskordade arvu patsiendi kohta. Esitatakse selgitused ja põhjendused teenuse arvu prognoosi kohta.

Teenuse näidustus	**Teenuse maht aastal *t***	**Teenuse maht aastal *t+1***	**Teenuse maht aastal *t +2***	**Teenuse maht aastal *t +3***
1	**2**	**3**	**4**	**5**
Multidistsiplaanrse meeskonna vastuvõtt parima ravi- jälgimisplaani koostamiseks	*130 tundi lastele* *130 tundi täisealistele*	*130 tundi lastele* *130 tundi täisealistele*	*135 tundi lastele* *135 tundi täisealistele*	*135 tundi lastele* *135 tundi täisealistele*
	260 tundi	260 tundi	270 tundi	270 tundi

6. **Taotletava teenuse kirjeldus**

6.1. teenuse osutamiseks vajalik koht (palat, protseduuride tuba, operatsioonituba, vm);

Vajalikud tingimused : ruum spetsialistide ja perega kohtumiseks, ambulatoorse vastuvõtu kabineti mööbel, koridor või koht kõnnirütmi hindamiseks, infotehnoloogilised vajadused (arvutid , internetiühendus, video ja fotojäädvustus tehnika)

6.2. patsiendi ettevalmistamine ja selleks vajalikud toimingud: premedikatsioon, desinfektsioon või muu;

Ettevalmistavaid meetmeid ei ole vaja

teenuse osutamise kirjeldus tegevuste lõikes;

NTD (neuraaltoru defekti alla liigituva harvikhaiguste patoloogiaga haigete grupp) **EKJP** (elukestva kroonilise jälgimise polikliiniku) **MDM** (multidistsiplinaarse meeskonna) vastuvõtu e. teenus skemaatiliselt : ARST neuroloog – ravijuht, ÕDE – koordinaa-

tor, ERIARSTID ja SPETSIALISTID (neuro)kirurg, ortopeed, nefroloog(uroloog), psühhiaater, füsioterapeut, psühholoog, sotsiaaltöötaja, logopeed(eripedagoog), silmaarst ja kurgu-kõrva-ninaarst vm spetsialist kaastakse vastavalt vajadusele

NTD EKJP- elukestvalt krooniliselt haigete jälgimise polikliiniku meeskond:
arst -/neuroloog, kirurg või internist/ – ravijuht (juhtumikorraldaja)
õde - koordinaator, arsti abiline,
eriarst id - (neuro)kirurg, ortopeed, nefroloog(uroloog), arst, keda kaastakse vastavalt vajadusele nagu psühhiaater, silmaarst, kõrva-nina-kurguarst, endokrinoloog, kardioloog,
spetsialistid - füsioterapeut, psühholoog, sotsiaaltöötaja, ortoosimeister, logopeed(eripedagoog), sotsiaaltöötaja

ARST – on kogu üksuse juht ja haige jaoks meditsiinilise tervisloo suunaja-hindaja, krooniliselt haige järjepidevuse hoidja meditsiinilises liinis

6.3. teenuse osutamise kirjeldus tegevuste lõikes;
Esitatakse taotletava teenuse osutamisel tehtavate tegevuste loetelu nende esinemise järjekorras, nende kirjeldus ning võimalusel lisada ka tegevuste sooritamise keskmised ajad.
6.4.

7. **Nõuded teenuse osutajale**

7.1. teenuse osutaja a, keskhaigla, üldhaigla, perearst, vm);
Teenuse osutajad on piirkondlikud kõrgema etapi haiglad (Tallinna Lastehaigla, edaspidi teenuse laienedes täiskasvanutele lisandub Põhja – Eesti Regionaalhaigla, Tartu Ülikooli Kliinikum). Kõrgema etapi raviasutus, kus on paljuprofiilse

ravi ja diagnostika võimalused ja kuhu on koondatud harvikhaigete hulk ning on omandatud kogemused-oskused erispetsiifiliste haigete ravimiseks ning dispanseerseks jälgimiseks

7.2. infrastruktuur, tervishoiuteenuse osutaja täiendavate osakondade/teenistuste olemasolu vajadus;
Tipphaigla, kus on võimalused kõrgtehnoloogiliseks diagnostikaks ja aparatuur lastekirurgia anestesioloogia, neurokirurgia jne, distsipliinide tööks ning eriarstid ja mehitatud abistruktuurid

7.3. väljaõppe vajadus;
Kaasasündinud väärarenguga sündinud laste ja edaspidi nendest kasvanud täiskasvanute interdistsiplinaarne ravi on pidevas arengus. Eriarstide ja erisptetsialistide osas on vajalik tetatud kitsam spetsialiseerumine

7.4. koolitused
Toimvad regulaarselt jooksvalt täiendõppekorras rahvusvahelistel seminaridel, konverentsidel, spetsialistide täiendõppe korras Euroopas (maailmas) asuvates vastavates keskustes.

7.5. minimaalsed teenuse osutamise mahud kvaliteetse teenuse osutamise tagamiseks;
Epidemioloogilisi andmeid arvestades, on minimaalne teenuse maht 125 tundi aastas, tagamaks jätkusuutlik haigete ravi ja jälgimine. Tegemist on harvikhaigete grupiga, kelle juurdekasv on aeglane ja praegu Eestis 137 SpB haiget ja 231 HC haiget. Seega haigete koondamine on hädavajalik arvestatava ravikvaliteedi tagamiseks ja haigete elukestva jälgimise korraldamise juurde

7.6. teenuse osutaja valmisoleku võimalik mõju ravi tulemustele.
Piirkondlikul haiglal peab olema vastava ettevalmistuse ja kompetentsiga spetsialistide olemasolu või väljakutsumise võimalus teisest pädevast raviasutusest

8. **Kulutõhusus**

8.1. teenuse hind; hinna põhjendus/selgitused;

MDM teenushinna baasmudel on koostatud vastavalt EHK teenuste ja uue teenuse vajadusi arvestades.

- EHK eriarsti 20 min. vastuvõtt kood 3002 maksab = 15,32 EUR-i
  60 minuti vastuvõtu hinnaks ühel arsti kohta = 45,96 EUR-i

MDM teenus eeldab võimalust tasustada 3 eriarsti s.o arsti komponent teenuses on seega **137,88 EUR-i**

o EHK õe 20 min. vastuvõtt kood 3035 maksab = 4,79 EUR-i 60 min. vastuvõtu hinnaks ühe õe kohta = 14,37 EUR-i Vastuvõtul on häireteta kulgemiseks vajalik hõlmata 2 õe haridusega liiget, seega õe teenuskomponent on **28,47 EUR-i** o Erispetsialisti teenuskomponent MDM vastuvõtul tuletub EHK füsioterapeudi teenusest kood 7050 = 10,57 30 min. 60 min. vastuvõtu hinnaks ühe füsioterapeudi kohta tuleb 21,14 EUR-i Vastuvõtul osaleb minimaalselt 2 erispetsialisti , kellede teenuskomponent on kokku **42,28 EUR-i** o Sekretär – asjaajaja, sotsiaaltöötaja tunnihind on MDM teenuse baasmudeli juurde arvestatud sotsiaalse rehabilitatsiooniteenuse arvestuslikust teenustunni hinnast **25,34 EUR-i** **Spetsialistide komplekshind kokku = 259,31 EUR-i** **Arvuti, telefoni, paberi jm kulu 4%spetsialistide kulus t = 10,37 EUR-i** **MDM vastuvõtu komplekshind on ümardamisega 270.-EUR-i**
8.2. teenuse osutamisega kaasnevate teenuste ja soodusravimite, mis on nimetatud p 4.4, isiku kohta kuni vajaduse lõppemiseni esitatud kulude võrdlus alternatiivsete teenuste kuludega isiku kohta kuni vajaduse lõppemiseni; Vajadusel täiendavate spetsialistide osalemine meeskonna vastuvõtul (ambulatoorse vastuvõtu hind)
8.3. ajutise töövõimetuse hüvitise kulude muutus ühe raviepisoodi kohta tuginedes tõenduspõhistele uuringutele võrreldes alternatiivsete teenustega;
8.4. patsiendi poolt tehtavad kulutused võrreldes alternatiivsete teenuste korral tehtavate kuludega

9. **Omaosalus**
9.1. hinnang patsientide valmisolekule tasuda ise teenuse eest osaliselt või täielikult *Patsientidel ja nende perekondadel puuduvad enamasti võimalused teenuse eest tasuda*

10. **Esitamise kuupäev**	*Märgitakse taotluse esitamise kuupäev järgmiselt: päev.kuu.aasta*
11. **Esitaja nimi ja allkiri**	*Allkirjastab taotluse esitanud erialaseltsi või - ühenduse, tervishoiuteenuse osutajate ühenduse või haigekassa juhatuse esimees/president või juhatuse liige. Elektroonsel esitamisel*

	allkirjastatakse digitaalselt ning lisatakse nime alla järgmine tekst "(allkirjastatud digitaalselt)".

12. **Kasutatud kirjandus**

1.*Journal of Pediatric Rehabilitation Medicine: AN INTERDISCIPLINARY APPROACH* ***Tase A***
Special isuse: Spina Bifida, Part 1
Guest editors: Tim Brei and Sue Mukherjee

LAST NUMBER OF THIS VOLUME
Editor – in – Chief: J.A.Neufeld *Volume 1, Number 4, 2008.*

2. *IF SpBH poolt korraldatud konverents Euroopa väikeriikide esindajatele Ghentis „The Turning Points conference“* ***Tase A***
02.02. – 04.02.2017. Lieven Bauwens`i –IF SpBH peasekretär - ettekanne 03.02.2017.

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twitter: @ifsbh | @PUSHtogether
facebook: www.facebook.com/ifsbh | www.facebook.com/push

3. © *1997 Rapid Science Publishers Quality of Life Research*, Vol **6**, 1997. pp. 123–132
This study was supported by a grant from the Ontario Ministry of Health (grant no. 02943) and from Bloorview Children's Hospital Foundation.

Development of a health-related quality of life instrument for use in children with spina bifida **Tase A**

P. C. Parkin,* H. M. Kirpalani, P. L. Rosenbaum,
D. L. Fehlings, A. Van Nie, A. R. Willan and D. King
Department of Pediatrics, University of Toronto, Toronto, Ontario, Canada
(P. C. Parkin, D. L. Fehlings); Departments of Pediatrics (H. M. Kirpalani,
P. L. Rosenbaum) and Clinical Epidemiology and Biostatistics (A. Van Nie,
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The objective of this study was to develop a spina bifida health-related quality of life (HRQOL) instrument. Items were generated through semistructured interviews, and reduced by frequency–importance product ranking. Validity was assessed by correlating the HRQOL score with a global question concerning the child's well-being using the Spearman's rank coefficient, and the Piers-Harris Children's Self-Concept Scale (P-H) using the Pearson correlation coefficient. Reproducibility was assessed at 2-week intervals using the intra-class correlation coefficient (ICC). Field testing was undertaken in a larger sample to evaluate item–total correlation, internal consistency and construct validity. Patients taking part in the study were 329 children and adolescents with spina bifida attending two treatment centres. Over 600 items were generated. These were reduced to 47 questions and 50 questions, for children and adolescents respectively. The correlation between the HRQOL score and the global question was r = 0.57, and with the P-H was 0.26 (children). These values for adolescents were 0.63, and 0.89, respectively. Reproducibility was ICC = 0.78 (children) and 0.96 (adolescents). Following field testing, the questionnaire was further reduced to 44 questions (children) and 47 questions (adolescents) by eliminating questions with an item– total correlation less than 0.20. Cronbach's alphas for the final instrument were 0.93 (children) and 0.94 (adolescents), and construct validity correlations were 0.63 (children) and 0.37 (adolescents). The spina bifida HRQOL instrument has good measurement properties and may be used as a discriminative instrument. Assessment of responsiveness is necessary before using it to evaluate therapy in clinical trials.

Key words: Health-related quality of life; pediatrics; spina bifida.

Introduction

Spina bifida is the most common disabling congenital anomaly with an incidence ranging from approximately 1.6–4 per 1,000 births in Canada.1 Recent advances have been made in the prevention of spina bifida through the administration of periconceptual maternal folic acid 2 and in prenatal diagnosis using maternal serum and amniotic fluid alpha-feto protein, and foetal ultrasonography.3 However, while prevention of disabling malformations is an important goal, individuals with spina bifida often experience considerable medical and psychosocial morbidity.

A number of longitudinal studies have assessed long-term outcome of individuals with spina bifida with respect to physical function, cognitive ability,4,5 psychosocial development 6,7 and the impact on the family.8,9,10 These studies suggest that spina bifida has a significant impact on the health of individuals, as is broadly defined by the concept of health-related quality of life (HRQOL).11,12However, it has also been shown that children and their families do not view their health as negatively as do their physicians.8,10 This suggests that HRQOL might best be understood

from the viewpoint of the individuals themselves, rather than from the viewpoint of clinicians. We wished to develop a HRQOL instrument in order to assess the specific question: what factors might predict long-term HRQOL in children and adolescents with spina bifida? Two approaches to

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10.PatrickDL,DeyoRA.Genericanddisease-specificmeasures in assessing health status and quality of life. *Med Care* 1989; **27**: S217–S232.
11.Juniper EF, Guyatt GH, Feeny DH, Ferrie PJ, Griffith LE,
Townsend M. Measuring quality of life in children with *Health-related quality of life in spina bifida*
Quality of Life Research . Vol 6 . 1997 **131 A**sthma. *Qual Life Res* 1996; **5**: 35–46.

4. The 58th Annual Meeting of the Society for Research into Hydrocephalus and Spina bifida Tase D

SRHS on ülemaailmne Spina Bifida ja Hydrocephaluse paoloogiaga tegelevte arstide, erialaspetsialistide, teadusuuringute, meditsiinitehnika arendajate jt. erialainimeste teadusühing, kes peab igal aastal temaatilist aastakoosolekut, kus esitletakse uurimistulemusi,ellu viidud uurimisprojekte originaalis ning uusimat erialalist infot-teavet kõige värskema esitlusena . Ühingusse saab astuda erialaarst /spetsialist isikliku sooviavalduse alusel ühenduse juhtkomiteele ja 2 juba ühenduses oleva erialaselt tunnustatud nõusoleva soovitaja toetusel. Reeglina ühenduse liikmeks kutsutakse. SRHS on SpBH patoloogia alal kõige innovatiivsem ja olukorrast maailmas teadlikum erialaste probleemide , uuringute , arengupoliitikat suunavate oma ala tippspetsialistide kogu.

5. **Tase D**

SKA ekspertarstidele loeng 05.05.2017. teemal „ Piirangud seljaajusonga ja šuntsõltuva vesipea korral“

https://www.dropbox.com/s/k7ipn9yuplhw6ga/ETTEKANNE%20SKA%20EKSPERTARSTIDELE%2005.05.2017.%20TLN-s.pptx?dl=0

Ettekanne sisaldab kõige värskemat statistikat NTD populatsiooni kohta Eestis seisuga 01.01. 2017.

- Haigete jaotumuses üle Eesti prevaleerub Tallinn koos Harjumaaga, järgnevad ligi 8x väiksemalt Ida-Virumaa, Lääne-Virumaa ja Pärnumaa.
- SpB haigeid 136 ja HC haigeid 231.
- Neurogeense põie dgn-ga lapsi 49, kellest 25 vajab elulistel näidustustel igapäevaselt IK-d
- Neurogeense põie dgn-ga täiskasvanuid 78, kellest igapäevaselt elulistel näidustustel vajab IK-d 31.
- Ratastoolis on SpB patoloogiaga haigetest 8 last ja 30 täiskasvanut

6. ROUNDTABLE MULTIPROFILE TEAM WORK WITH SpBH PATIENTS IN THE CONTEXT OF THE MODERN MEDICINE **Tase D**
03 – 04 December 2015, Tallinn, Estonia, Tallinn Children`s Hospital in Mental Health Centre (Tervise 28)

7. 17.wissenschaftliche Tagung des ASBH Beirats 20. – 21. 11.2015. in Fulda **Tase D**
www.bonifaciushaus.de
Ülesaksamaaliselt Seljaajusonga ja Vesipeahaigete Keskus ning kõiki Saksamaal elavate SpBH haigeid ühendav ühing ASBH korraldab regulaarselt igal aastal teematilise „päevakajalise“ õppe-koolitus ja ülevaate seminari, mis on kõigi saksa keelsete riikide SpBH haigete ja nende ravimisega tegelevate arstide, spetsialistide, tervishoiukorraldajate jaoks oluline aasta tähtsündmus

8. Amtsgericht Dortmund HRB 26271

Gesellschaft der Arbeitsgemeinschaft Spina Bifida und Hydrocephalus e. V. (ASBH)
www.asbh.de
Facebook: https://www.facebook.com/asbhdeutschland

Multimediabroschüre
http://www.unserebroschuere.de/ASBH/MailView/

Selbständigkeitstraining, Schulungen, Tagungen und lohnende Veranstaltungen
www.asbh.de/termine/index.html **Tase A**

Ratgeber zu Spina bifida und Hydrocephalus
www.asbh.de/asbhratgeber/asbhratgeber.html **Tase A**

Fachbeiträge
www.asbh.de/fachbeitraege/fachbeitraege.html **Tase A**

9. Carla Verpoorten – Belgia Ghenti Ülikooli lasteneuroloog`i loeng IF SpBH seminaril 03.02.2017. **Tase D**

10. Guidelines for Spina Bifida Health Care Services Throughout the Lifespan. Council PA. In: Merkens MJ, ed. *version 3*: Spina Bifida Association; 2006:70. **Tase A**

Overview of the Health Care Guidelines for Spina Bifida Across the Lifespan

Brad Dicianno, MD
Co-Chair, SB Healthcare Guidelines Steering Committee
Chair, Professional Advisory Council at SBA
Associate Medical Director for Adult Care at SBA

SPINA BIFIDA WORLD CONGRESS

1)

Disclosures

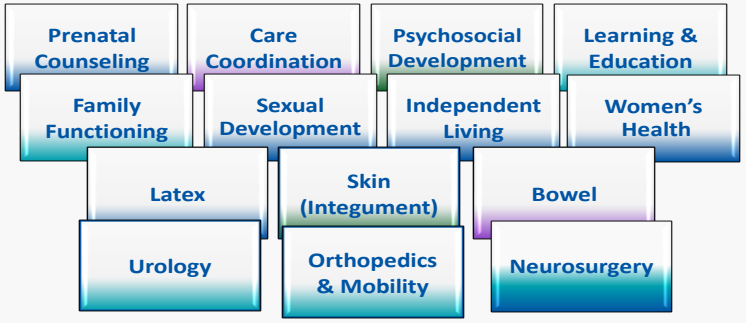
- No conflicts of interest to report.
- This project is supported by:
  - The Spina Bifida Association
  - Spina Bifida Collaborative Care Network
  - The Centers for Disease Control and Prevention

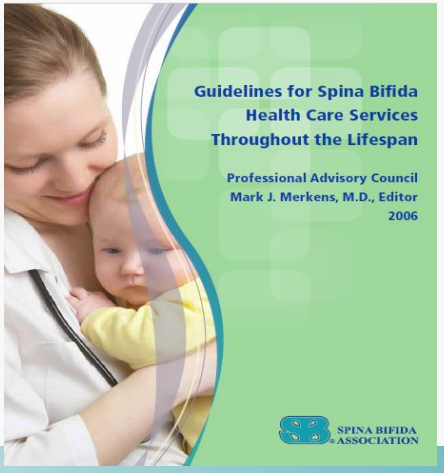
SPINA BIFIDA WORLD CONGRESS

2)

Guidelines for Spina Bifida Health Care Services

- Prior guidelines published in 2006



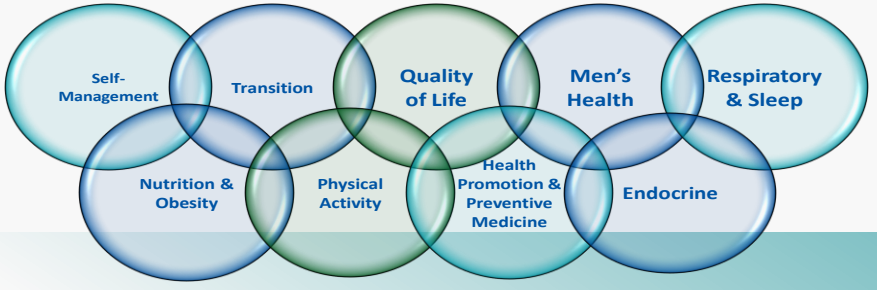


SPINA BIFIDA WORLD CONGRESS

3)

Overview of the New Guidelines

- 2014 -- SBA and CDC convened steering committee as part of SBCCN
- Concluded an update and expansion needed
- 9 new topic areas suggested:



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4)

Methodology

- Recognized consensus-building methodologies for the development of clinical practice guidelines
  - Single Text Procedure
  - Nominal Group Technique
- More than 80 experts participated in the process
- **Multi-disciplinary groups**
- Spans newborn to adult

SPINA BIFIDA WORLD CONGRESS

5)

Format of New Healthcare Guidelines Document

- Outcomes – what we wish to achieve with good care
- Clinical questions – important questions that were targets of the lit review
- Guidelines – recommendations for care that are based on the lit review and clinical consensus
  - Metrics for good care
- Research gaps – important topics where research and clinical consensus are lacking
  - Synergistic areas with NSBPR

SPINA BIFIDA WORLD CONGRESS

6)

Conclusions

- Encourages evidence- and consensus-based practices
- Published and disseminated to the SB community
- Goal to optimize outcomes and improve care for all people living with SB

SPINA BIFIDA WORLD CONGRESS

7)

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SPINA BIFIDA WORLD CONGRESS

8)

11.

Second World Congress on Spina Bifida Research and Care 2012 . USA, Las Vegas **Tase A**

Urology Abstracts

1.Title: Medical and Surgical Management of Adult Patients with Spina Bifida

Presenting Author: Jessica Casey, MS, MD, Northwestern Memorial Hospital USA, Department of Urology

Additional Authors:

- Alyssa Greiman, BS, Northwestern Memorial Hospital, Department of Urology
- Kalen Rimar, BS, Northwestern Memorial Hospital, Department of Urology
- Stephanie Kielb, MD, Northwestern Memorial Hospital, Department of Urology

Background: Medical management of spina bifida patients is commonly complicated by their multiple medical co-morbidities, and there are relatively few studies describing their urologic management as adults. We present medication use and surgical management of patients from our adult spina bifida clinic.

Methods: A retrospective review of patients seen in our adult spina bifida clinic from January 2004 to November 2011 with diagnosis of myelomeningocele, spina bifida occulta, and tethered cord syndrome was performed. Urologic medications and surgeries are reported.

Results: Our clinic consists of 229 patients; 134 women (58.5%) and 95 men (41.5%) average age 31years (range: 19-79, SD 9.26), with average age of first visit 27 years (range: 16-72, SD 9.13). 111 patients (48.5%) are on anticholinergic medications. The majority (84 patients, 75.7%) were on one anticholinergic medication and 23 patients (20.7%) were on two. 58 patients are on oxybutynin, 47 solifenacin, 24 darifenacin, and 7 on tolterodine. Twelve patients are using gentamicin installations to manage recurrent infections. 188 (82.1%) underwent at least one urologic surgical procedure including augmentation cystoplasty (29 12.7%) and ileal conduit (9 patients, 3.9%). 35 patients (15.3%) had a procedure for stone disease. 33 (14.4%) under went a continence procedure including pubovaginal sling (14 patients, 42.4%) botox injection (6 patients, 18.2%), collagen injection (5 patients, 15.2%) artificial urinary sphincter (5 patients, 15.2%), and male Stamey slings (3 patients, 9.0%). Only 2 patients (6.1%) had continence procedures performed before the age of 18.

Conclusion: Most adult spina bifida patients continue on anticholinergic medications and most have undergone at least one urologic surgical procedure, augmentation cystoplasty and stone treatment being the most common. Pubovaginal sling placement was the most common operative procedure for urinary incontinence; most continence procedures were performed **after age 18**.

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Urology Abstracts

2.Title: Variability in Bowel and Bladder Incontinence Across US Spina Bifida Programs

Presenting Author: Heidi Castillo, Dr., Cincinnati Children's Hospital Medical Center

Additional Authors:

- Jonathan Castillo, Dr., Cincinnati Children's Hospital Medical Center
- Minn Soe, Dr., Centers for Disease Control and Prevention
- Kevin Liu, Dr., Centers for Disease Control and Prevention
- Beth Ward, Dr., Centers for Disease Control and Prevention
- Mark Ward, Dr., Centers for Disease Control and Prevention
- Michael Schechter, Dr., Emory University School of Medicine

Background: Although life expectancy has dramatically increased over the last half century, **many secondary impairments and barriers to full participation in life's activities remain for people with Spina Bifida (SB). Bowel and bladder incontinence** is one such impairment. This study's purpose was to examine differences in percent bowel and bladder continence across **SB clinics in the U.S. that contribute clinical and demographic information to the National Spina Bifida Patient Registry (NSBPR).**

Methods: Data on continence and its management was collected through the NSBPR from patients receiving care in one of nine SB centers across the US from March 2009 to April 2011. Continence was defined as no involuntary stool leakage or dry, with or without interventions, during the day. We first conducted descriptive statistics and examined differential distributions of bowel and bladder continence by individual patient characteristics. We then conducted multivariable logistic regression analysis to model continence by clinic sites after adjusting for differences in the distribution of patient characteristics.

Results: Among the study participants, 1,041 patients were 5 years of age or older. Nearly 40% and 35% of participants with myelomeningocele were continent of stool and urine respectively. There was significant differential distributions of continence by individual patient characteristics such as age at first annual visit, sex, race/ethnicity, health insurance, level of lesion, SB type, and method of mobility and clinic sites (all p-values<0.05). After controlling for these characteristics, the adjusted percentage of continence among the nine study sites ranged between 4.1% and 62.6% for bowel and 18.7% and 52.6% for bladder.

Conclusion: Synchronous data collection on bowel and bladder continence and management from patients receiving care across the US is feasible through the NSBPR. Among this population, there was notable variability in the percentage of continence. Further analysis of attributable factors is indicated to appraise the documented variation in continence outcome. **NB**! Rahvuslik SpBH register

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Urology Abstracts

3.Title: Urinary Continence in Young Adults: Perceptions, Reality, and Limitations

Presenting Author: Courtney Shepard, MD, Children's Hospital of Alabama

Additional Authors:
- David B. Joseph, MD, Children's Hospital of Alabama

Background: As patients with spina bifida approach young adulthood, the urologist's role expands from preserving the upper tracts to assisting with continence of urine and stool. However, continence in this population remains poorly defined. We review our spina bifida

population to determine who is continent, who is working on continence, and who has opted out of a continence program. We try to identify factors that promote continence with a secondary goal of assessing the effectiveness of our registry to answer these questions.
Methods: 337 patients with spina bifida aged 3 months to 29 years participate in our IRB approved National Spina Bifida Patient Registry and EMR. We assessed the age, continence status (defined as no more than 1 incontinent episode in the past 6 months), bladder and bowel management and surgical history of these patients. We reviewed the EMR to determine the current treatment program and if the patient is working on continence or has been reconstructed. Factors that have led to failure are assessed as are factors that have led to acceptance of incontinence without a treatment program.
Results: 115 patients were < 7 years of age, 91 were 7-12, 81 were 13-17, and 50 were >18. Of those <7, 10.4% were continent of urine and 9.6% continent of stool. Patients 7-12, 31.9% were continent of urine and 35.3% were continent of stool. Patients 13-17, 40.7% were continent of urine and 29.6% continent of stool. Patients > 18, 48% were continent of urine and 50% continent of stool. Continence and associated treatment factors are detailed in each age group. Limitations of our registry are explored.
Conclusion: Continence is considered important for social acceptance. Despite this, continence remains low in our study group. Contributing factors include level of dependent care, satisfaction with current level of continence, no desire for surgery, and lack of perceived improvement of lifestyle. Concrete assessment of continence is needed along with validated assessment tools to determine the true impact of incontinence in the spina bifida population.

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Urology Abstracts

4. Title: ***Refinement of Continence Questions for the National Spina Bifida Registry***

Presenting Author: Kathryn Smith, RN, DrPH, Children's Hospital of Los Angeles
Additional Authors:

- Karen Rauen, RN, MSN, Children's Hospital of Wisconsin
- William O. Walker, Jr, MD, University of Washingotn School of Medicine; Chief, Division of Developmental Medicine, Seattle Children's Hospital
- Judy Thibadeau, RN, MN, Centers for Disease Control and Prevention
- Elizabeth Ward, RN, MPH, Centers for Disease Control and Prevention

Background: There are no standards for defining or quantifying bladder or bowel continence in patients with spina bifida. As part of the National Spina Bifida Registry, questions are asked about whether the patient is continent of urine, defined as ?dry, with or without interventions during the day? and continent of stool, defined as ?no involuntary stool leakage, with or without interventions, during the day.? Additional information about continence is needed to understand the impact on the daily lives of individuals with spina bifida, as well as to determine the effectiveness of various interventions and the need for changes.
Methods: A small group of expert clinicians representing sites participating in the National Spina Bifida Registry, as well as staff from the Division of Human Development and Disability, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention **convened by phone several times to determine additional questions** to be added to the Registry. Discussion occurred around the benefits of each additional element and consensus was achieved before adding any questions. Subsequently, the process and questions were reviewed with the Registry's Coordinating Committee for general approval

Results: An additional three questions were added to the Registry related to both bladder and bowel continence, including the frequency of incontinence, bladder and bowel care interventions, and self-care. More choices were also added for clarification within the questions.
Conclusion: A small group process is reasonable for identifying additional questions to be included in the **National Spina Bifida Registry**, using a representative group of expert clinicians. More detail will satisfy research queries as well as help clinicians evaluate the outcomes of their interventions.
NB! **National Spina Bifida Registry** tähtsus

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Urology Abstracts

5.Title: Long-Term Follow-Up of Incontinent Upper Tract Urinary Diversions (UD) in Spina Bifida Patients

Presenting Author: Sherry Ross, MD, Division of Urology, Department of Surgery, Duke University Medical Center
Additional Authors:

- John Wiener, MD, Division of Urology, Department of Surgery, Duke University Medical Center

Background: Prior to widespread acceptance of clean intermittent catheterization, SB patients at high risk for upper tract damage were managed with incontinent UD. Although UD is now rarely used, many SB patients who received UD as children are still alive today, but little is known about the long½'term sequelae.
Methods: Review of 418 patients followed at our SB clinic was performed to identify those >21y of age who have incontinent upper tract UD. Data were reviewed to assess renal function, incidence and management of nephrolithiasis, urinary tract infections (UTI), metabolic disturbances, post½'surgical complications and need for surgical revision, and development of malignancy.
Results:
We identified 9 SB patients who had undergone incontinent upper tract UD (7 ileal loop diversion, 1 colonic loop diversion, 1 cutaneous ureterostomy). Mean follow½'up was 29.7 years (range 9- 40 years). There has been no known disease related mortality. The most common complication was UTI (8/9 patients) with 5/8 patients requiring 1 admission for urosepsis and/or pyelonephritis. Recurrent nephrolithiasis was noted in 7/9 patients with 6/7 patients undergoing 2½'9 stone½'related procedures.
Remarkably, although 7/9 patients have evidence of renal atrophy on imaging, chronic renal insufficiency has only developed in 1 patient with medical renal disease (serum creatinine of 2.3 mg/dL.) Only 1/9 patients has developed hypertension. Loop revisions were performed in 2/9 patients while peristomal hernias were identified in 3/9 patients with only 1 patient requiring surgery. Undiversion was undertaken in 2/9 patients, although both patients ultimately reverted to incontinent UD. Neither chronic metabolic abnormalities nor malignancy within the GU tract were noted.
Conclusion:
Long-term follow up of SB patients with incontinent upper tract UD revealed good renal preservation. Not surprisingly, delayed post operative complications were noted in 100% of patients. The most common complications were UTI and nephrolithiasis, and the majority has required surgical management of complications.

6.Title: Early Initiation of CIC does not Prevent Renal Scarring in Patients with Neurogenic Bladder and a History of VUR
Presenting Author: George Chiang, MD, UCSD Pediatric Urology
Additional Authors:

- George Kaplan, MD, UCSD Pediatric Urology
- Kerrin Palazzi-Churas, MPH, UCSD Urology

Background: Clean intermittent catheterization (CIC) has been reported to resolve vesicoureteral reflux (VUR) in a majority of patients with neurogenic bladder. Renal scarring is highly correlated with the presence of VUR. We examined the risk factors for DMSA abnormalities in the context of VUR in a population where age at CIC initiation was not consistent.
Methods: A retrospective chart review was performed of all patients presenting to the spinal defects clinic at Rady Children's Hospital from June 2007 to October 2011. Of 292 patients, 100 patients underwent DMSA scans. Variables recorded included: spinal lesion, VUR history (Grade 1 or greater), hydronephrosis (Grade 1 or greater), UTIs, DMSA indication, the presence of photopenic defects, the most recent urodynamics results, and age at initiation of CIC.
Results: Photopenic defects were present in 43% of all patients and a history of VUR was present in 32%. 66% of patients with a history of VUR had evidence of DMSA abnormalities. 54 patients started CIC after birth (median age 5) with 30% of patients having a history of VUR. Photopenic defects were found in 9/16 (56%). 17 patients started CIC at birth with 48% having a history of VUR. Photopenic defects were found in 7/8 (88%). On multivariate analysis, a history of VUR (OR 4.4 p=0.015), hydronephrosis (OR 5.1 p=0.008) and CIC at birth (OR 4.4 p=0.03) were all significantly associated with photopenic defects. No association was seen between age at CIC and sex for predicting photopenic defects.
Conclusion: Early initiation of CIC does not always prevent the deleterious effects of the neurogenic bladder. **Hydronephrosis and VUR have a high possibility of causing renal scarring. More aggressive initial measures may be required in this high-risk population.**

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Urology Abstracts

7.Title: Distribution, Care and Management of Spina Bifida in Kenya
Presenting Author: Gladys Tallam
Methods:

- General distribution of people living with spina bifida.
- Beliefs about *spina bifida*

- Challenges facing parents as they bring up their kids in Kenya. Many children below 7 years are still alive, but the percentage of those who are above 7 years decreases as their parents get demoralized when they notice that their children do not progress in these ways:
- The child fails to walk independently
- The child fails to have sensitivity in some parts of the body.
- The lack of sensitivity may also cause ulcerations.
- The child and the family may lack awareness and knowledge about spina bifida.
- Barriers to inclusive education in our country; for example, lack of well-sanitized area for catheterization for those with bladder and bowel incontinence.
- Our hopes and dreams as Bethany Kids in raising awareness about this condition.

Conclusion:

- Many children who walk have difficulty holding both their bladder and their bowel. I assume that their nerves 'concentrate' on walking rather than holding the bladder and bowel. They use oxybutinin to help hold the bladder. Many of these children suddenly come to the hospital with deep debicuters ulcers when it is too late and suddenly die because of the incurable ulcers.
- Those who use crutches have a better capacity of holding urine as compared to those who walk (that is my assumption). They can hold without using drugs. As long as they continue doing CIC, then they will be free from ulcers and kidney problems.
- Patients on wheelchairs experience difficulty in emptying their bowel but can empty the bladder well. They experience constipation and urine retention. Failure to empty the bladder results in hydronephrosis.

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Urology Abstracts

8.Title: Cost Savings from not Catheterizing Newborns with Spina Bifida

Presenting Author: Candace F. Granberg, MD, Children's Medical Center/UT-Southwestern Medical Center

Additional Authors:

- Warren T. Snodgrass, MD, Children's Medical Center/UT-Southwestern Medical Center
- Micah A. Jacobs, MD, Children's Medical Center/UT-Southwestern Medical Center

- Patricio C. Gargollo, MD, Children's Medical Center/UT-Southwestern Medical Center

Background: One published option for urologic management of newborns with spina bifida is universal therapy, placing all patients on clean intermittent catheterization (CIC) and anticholinergic medication (AC). An alternative is selective therapy based on initial urodynamic studies (UDS), placing those with end-fill pressures (EFP) > 40cmH2O on CIC+AC.
Cost savings of selective therapy were calculated.
Methods: We prospectively managed infants with spina bifida based on initial UDS at age < 6mo. Those with EFP<40cmH2O were observed, while those with EFP>40 started CIC+AC. Follow-up UDS were performed at age 1 year or for clinical indications (new hydronephrosis, recurrent febrile UTI). Observed patients demonstrating new EFP>40cmH2O started CIC+AC. Cost savings from not performing CIC+AC from birth until age 3 years, when therapy for social continence often begins, were calculated.
Results: 61 infants (30F:31M) had minimum follow"up 12mo (median 41); 12 (20%) with EFP>40 were assigned CIC+AC. 6/49 (12%) observed patients developed new EFP>40 at median age 9 mo and began CIC+AC. Therefore, 18/61 (30%) began CIC+AC by age 1 year. 70% did not begin CIC+AC until age ½ ½3 years for continence. Costs incurred in those 70% had they been treated with CIC+AC since birth were calculated. Using Texas Medicaid reimbursement rates, annual savings in one patient from not performing CIC was $4363.20 ($2.02/catheter x CIC 6x/day x 30 days x 12mo), and for not prescribing AC (0.2mg/kg BID) was $480 ($40/month x 12mo). Total cost savings for not performing CIC+AC for one year in one child was $4843.20 ($14,529.60 for 3 years). Savings for 43 patients was $624,772.80.
Conclusion: 20% of newborns had EFP >40 on initial UDS, and 12% of observed patients developed EFP >40 warranting CIC+AC. 70% had no UDS or clinical indication for therapy until desire for continence. Not performing CIC+AC in this group saved > $600,000. Selective therapy in newborns with spina bifida was clinically safe and cost-effective.

9.Title: Continence Management in Children and Young Adults with Spinal Cord Lesion in Developing Countries
Presenting Author: Myleen Christiaens, RN, University Leuven Belgium
Additional Authors:

- Gunnar Buyse, PhD , University Leuven Belgium
- Carla Verpoorten, MD, University Leuven Belgium

Background: This session will focus on continence management in children and young adults with spinal cord lesions in developing countries. The aim is to demonstrate that new insights in neurogenic bladder treatment and standards of good care in more developed countries, can be translated to patients and caregivers in developing countries.
Methods: Early detection of the infant at risk and early adequate treatment (clean intermittent catheterisation (CIC), oxybutynin), are the cornerstones of the management to achieve renal protection and social continence. CIC has been proved to be safe, even in newborns. The practice of CIC has dramatically reduced urinary tract infections. In addition, combined with oxybutynin if necessary, this

treatment prevents renal and bladder damage. Importantly, with CIC alone or if required combined with oxybutynin social continence may be achieved.
Results: In developing countries, recent information and training material on continence management are not readily available. We have developed a practical and feasible protocol for management of the
neurogenic bladder in developing countries, including easy teaching and training material. The
International Federation for Spina Bifida and Hydrocephalus, who promotes worldwide total care for
these children, has enabled the application of this protocol
Conclusion: Neurogenic incontinence is a devastating handicap with severe medical and social
implications, certainly in developing countries. The medical problems, causing high morbidity and
eventually mortality, start early in life long before the incontinence becomes a concern. The incontinence and its consequences have severe psychosocial implications, such as social isolation and often refusal at school. The continence **management program** of the International Federation for Spina Bifida and Hydrocephalus, opens the door for many children and adults for social inclusion.

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Urology Abstracts

10.Title: Chronic Kidney Disease in Children with Spina Bifida: An Under-recognized Co-morbidity
Presenting Author: Michelle Baum, MD, Children's Hospital, Harvard Medical School
Additional Authors:

- Deborah Stein, Pediatrics, Clinical Research Program, Harvard Medical School
- Catherine Gordon, Pediatrics, Clinical Research Program, Harvard Medical School
- Henry Feldman, Clinical Research Program, Harvard Medical School
- Nedda Hobbs, Pediatrics, Harvard Medical School
- Stuart Hobbs, Urology, Harvard Medical School

Background: Risk factors for chronic kidney disease (CKD) in children with spina bifida (SB) may
include neurogenic bladder, urinary obstruction, and vesicoureteral reflux, all of which lead to renal
scarring. The lifespan of children with SB has significantly increased and the prevalence of CKD
remains unknown. Serum creatinine may be a poor marker of renal function, given that creatinine
generation correlates with muscle mass. Thus, CKD may be undetected in some children if assessment is based purely on serum creatinine measurements. The objective of this study was to determine the
prevalence of CKD in this cohort and to define associated risk factors.
Methods: We conducted a retrospective chart review of patients in the Children's Hospital Boston
Myelodysplasia Program from 1999 to October 2011, age 1 month to 19 years old. Data on the first 250 subjects is presented (108 male, 142 female).
Results: Of the 197 subjects with lab data available, 29 (15%) had CKD as calculated by either an

estimated GFR using serum creatinine or as documented by a physician. Of the 248 with blood pressures documented, 27 (11%) had hypertension (HTN), and CKD was significantly associated with HTN ($p<0.001$). 72 had a DMSA renal scan and 44 (61%) had renal scarring. CKD was significantly
associated with renal scarring ($p=0.022$). The majority 196/227 (86%), had documented neurogenic
bladder dysfunction.
Conclusion: CKD is a common co-morbidity in children with SB, even when using serum creatinine as a marker of renal function. The high prevalence of renal scarring, HTN and neurogenic bladder in this population indicates a need for increased awareness and screening for CKD. CKD may be underestimated and thus undetected in children whose creatinine is not abnormal. More reliable methods of measuring renal function may be needed in children at risk for CKD.

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Urology Abstracts

11.Title: Characteristics and Survival of Persons with Spina Bifida (SB) and End Stage Renal Disease (ESRD) using Data from the United States Renal Data System
Presenting Author: Lijing Ouyang, PhD, Centers for Disease Control and Prevention
Additional Authors:

- Rodolfo Valdez, Centers for Disease Control and Prevention
- Judy Thibadeau, RN, MN, Centers for Disease Control and Prevention
- Julie Bolen

Background: Renal complications frequently cause long-term morbidity and mortality in persons with SB. ESRD is the most severe form of renal disease. The characteristics and mortality patterns of persons with SB who develop ESRD have not been examined in a US national sample.
Methods: We conducted a retrospective analysis using the United States Renal Data system (USRDS). ICD-9-CM code 741 was used to identify persons with SB from USRDS hospital inpatient data (1977 - 2007) and physician and facility claims (2004ˆ'2008). We compared demographic and ESRD-related characteristics between persons with and without SB, all of whom had ESRD. Kaplan-Meier and Cox proportional hazards survival analysis were performed for those who developed ESRD since 2004 and were followed until 2007.
Results: As of 12/31/2006, a total of 956 living persons were identified as having both SB and ESRD in the USRDS database, averaging 41 years old, compared to 615,508 persons with ESRD without SB who averaged 60 years old. The lead primary cause of ESRD is urologic conditions for persons with SB (35%) and diabetes for persons without SB (37%). Unadjusted analysis using Kaplan-Meier curves demonstrates a survival advantage of persons with SB compared to persons without SB. Cox proportional hazards
model adjusting for sex, race, age, and primary cause of ESRD showed that the survival advantage of
persons with SB appeared to be associated with younger ages at onset of ESRD.
Conclusion: Our study demonstrates that having SB does not significantly increase the mortality hazard for patients with ESRD. However, since persons with SB appear to develop ESRD at a much younger age than persons without SB, this suggests the need to monitor renal function more aggressively in young adults with SB. **If quality urologic care is provided to persons with SB, they should transition through life with normal renal function**.

12.

Second World Congress on Spina Bifida Research and Care 2012 . USA, Las Vegas **Tase A**

Psychosocial/Quality of Life Abstracts

1.Title: Children with Spina Bifida in Mongolia

Presenting Author: Ariunsanaa Batsaikhan, Maral-Elch Foundation

Additional Authors:

- Enkhchuluun Rinchin, Maral-Elch Foundation

Background: Mongolia is a Northeast Asian country with a population of over 3.1 million. 59% of the population is under the age of 30 and from this 27% is under the age of 14. According to unofficial
statistics, there are around 34 to 37 thousand children in Mongolia who live with disabilities, 12% of
which have not been officially diagnosed, and 71% of the children do not get rehabilitation they need.
Recently, the number of children born with birth defects rose by an alarming number, especially infants with spina bifida.

Methods:

- At this point, the Mongolian government and the Ministry of Health do no hold any official
statistic of children with Spina Bifida.
- Majority of our research is based on hospital records
- Independent research findings
- Interviews with doctors

Results: Recently, the number of birth defect increased significantly, mostly dominated by cerebral palsy and spinal bifida. Infant birth defects jumped from 4.9% in 2004 to 8.3% in 2008. Nationally, there are only two hospitals that operate on children with spina bifida; one of them is Shastin Hospital. According to hospital records, the number of infants born with spina bifida increased six times, 49% of which die from complications. A handful of Mongolian doctors are able to operate on children with spina bifida but there are no rehabilitation programs available for the children. Also, there are no medical professionals focusing on spina bifida research and education. In Mongolia there is a significant lack of public knowledge concerning spina bifida.

Conclusion: Maral-Elch, Foundation to bring awareness for Mongolian Children with Spina Bifida, is the only organization in Mongolia representing children with spinal bifida and their families and we need support from international specialists and organizations. **Currently we are accepting all level of support from diagnosis to rehabilitation.**

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Psychosocial/Quality of Life Abstracts

2.Title: Change in Mobility in a Post-Pubertal Cohort with Spina Bifida

Presenting Author: Drew Davis, MD, University of Alabama at Birmingham/Children's of Alabama

Additional Authors:

- Anastasia Arynchyna, CHS Pediatric Neurosurgery
- Steven Veselsky, CHS Pediatric Neurosurgery

- Amita Bey, MPH, CHS Pediatric Neurosurgery
- Chevis Shannon, MBA, DrPH, Children's of Alabama/UAB

Background: Decline in mobility in individuals with spina bifida (SB) has been described in cohorts both pre- and post- puberty and has been ascribed to obesity, precocious puberty, age, tethered cord, and VP shunt failure, among others. While children with SB may have multiple factors contributing to their mobility status or change therein, there is a lack of conclusive data to inform discussion of prognosis and medical decision making. This review utilizes the recently created CDC SB database to evaluate change in mobility in a cohort with SB managed at Children's of Alabama.
Methods: The CDC database was queried with regard to a cohort age 17 (+/- 12 months), presumed to be post-pubertal. Lesion level, mobility status, and surgical history were assessed and then compared to the same data for each patient at all previous visits. A chart review was completed for those subjects for whom complete data had not been entered into the CDC database.
Results: 36 individuals age 17 (+/- 12 months) were identified. Of these 9 (25%) experienced a change in mobility status detected over the course 7 (+/- 2) clinic visits. Some experienced temporary changes that represented declines or improvements over time. There were no statistically significant differences between the group that experienced a change in mobility and those that did not.
Conclusion: Children and adolescents with SB may experience both declines and improvements in mobility over time. While multiple factors are believed to contribute to a change in mobility in
individuals with SB, none were clearly identified in this review. Further research regarding contributors to change in mobility status in SB is needed and should be evaluated in larger cohorts.
Elukestvalt muutustes

13.

Thirth World Congress on Spina Bifida Research and Care 2017 . USA, Coronado **Tase A**

1.Title: **Health and Function . The ICF Model**
Children`s Hospital of Pittsburgh of UPMS. Rehabilitation Institute
Author: Amy Houtrow, MD, PhD, MPH

ICF Classification definition of children's health:

- Develop and realize their potential
- Satisfy their needs
- Develop the capacities that allow them to interact successfully with their biological, physical, and social environments

Disability is an umbrella term for **impairments** /kahjustus/ at the level of the body part, **activity limitations** / tegevus piiratus, kitsenemine/ at level of person, and **participation restrictions** / osavõtu kitsendus, piirang/ at the level of the person in society
Disability is viewed as the outcome of the interaction between health conditions and contextual factors

14. „ The Groningen Protocol – Euthanasia in Severely III Newborns`" , Verhagen **Tase D**

352 The New England Journal of Medicine, 10 March (2005), 959.

Spina bifida is one of the most common congenital defects, occurring within the first 25 day of pregnancy. The spine does not clouse completely and the nervous system is frequently damaged with resulting paralysis. Most will have problems with bowel and bladder control. Other problems also arise. Hydrocephalus arises from the same causes and results in blockage of drainage pathways for fluids around the brain. The fluid accumulates in the brain, causing compression of surrouhding tissue. In most cases several surgeries are needed. Good medical care and intensive training for the person and family can prevent many complications and improve the person`s life. www.ifglobal.org

15.

Second World Congress on Spina Bifida Research and Care 2012 . USA, Las Vegas **Tase A**

Adult Care/Transition Abstracts

1.Title: ***From Transition Challenges to Successes: Establishing a Spina Bifida Adult Care Clinic***

Presenting Author: Betsy Hopson, BA, Children's Hopsital of Alabama

Additional Authors:

- Chevis Shannon-Waters, MBA, MPH, DrPH, Children's Hospital of Alabama, University of Alabama - Birmingham
- Richard Drew Davis, MD, Children's Hospital of Alabama, University of Alabama - Birmingham
- Jeffrey Blount, MD, Children's Hospital of Alabama, University of Alabama - Birmingham
- Patrick Pritchard, MD, University of Alabama - Birmingham

Background: We previously conducted a study to evaluate how patients were transitioned from pediatric and adolescent care to adult care. Our findings were presented at the First World Congress on Spina Bifida meeting. The purpose of our current study was to describe the challenges previously identified and to evaluate our success in providing a more comprehensive and collaborative adult care clinic.

Methods: In 2010 we established a comprehensive adult care clinic facilitating transitional care for many of our spina bifida patients. Benchmarks were developed based on previously identified challenges including: 1) the need to customize a goal-specific action plan for each individual participant based on his/her capabilities, and 2) the establishment of effective interaction between key service providers to facilitate referrals, develop criteria for transition and follow up visits.

Results: With the establishment of our adult care clinic we have developed a lifetime care model for approximately 60 patients with Spina Bifida using the CDC funded electronic medical record. Full

support and commitment of our medical director, including monthly participation in the adult clinic, established an environment for success. Additional neurosurgical resources were identified with both pediatric and adult neurosurgeons working together to ensure effective outpatient care and surgical intervention. The pediatric spina bifida coordinator oversees the adult clinic creating continuity of care, facilitating a care plan utilizing previous medical history and addressing unmet needs. Familiar faces and similar practice patterns allows our patients the opportunity to transition with less anxiety and a better understanding of their future medical care.

Conclusion: Collaborations between the pediatric and adult care providers have created an opportunity for successful transition of our spina bifida patients. We will continue to prospectively follow these patients to further determine challenges and successes including improved quality of life and patient satisfaction.

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Adult Care/Transition Abstracts

2.Title: *Implementing the Spina Bifida Registry with an Existing Legacy EMR: Trials, Tips and Solutions*

Presenting Author: Kurt Freeman, PhD, Oregon Health & Science University

Additional Authors:

- Daria Ettinger, Oregon Health & Science University
- Elizabeth Adams, Oregon Health & Science University
- Mark Merkens, Oregon Health & Science University

Background: The OHSU Spina Bifida Clinic has participated on the CDC-sponsored patient registry since its inception. Simultaneously, the CDC and SBA are promoting a Spina Bifida Electronic Medical Record (EMR), designed to assist clinics in standardizing documentation (and thus possibly care). Clinics participating in the registry project utilize the SB EMR for imputing and transmitting registry variable information. The existence of dual EMR systems (i.e., SB EMR and legacy EMR) can create unique challenges related to data extraction and transmission for participating in the SB registry project. The goal of this paper is to highlight those challenges and offer suggested solutions.

Methods: This presentation represents a descriptive assessment of experience attempting to implement the SB registry within a health care system that utilizes a legacy EMR.

Results: Through our experience with the registry project thus far, we have learned that data collection and transmission into the National Spina Bifida Registry is a complex and time consuming process. In an effort to reduce redundancy of data collection and registry submission, we have partnered with clinicians and our Information Technology Group to develop templates for data collection of registry data within OHSU's legacy EMR.

Conclusion: The OHSU experience with the registry, SB EMR, and legacy EMR highlights the importance of considering processes for involvement in the national registry project. Success requires

careful collaboration with clinicians to ensure consistent documentation of registry variables and workarounds for extraction of relevant information from medical charts, all while considering efficiency and sustainability.

3.Title: ***Life Course Health Patterns and Health Care Utilization among Adolescents and Adults with Spina Bifida***

Presenting Author: Gregory Liptak, MD, MPH, Upstate Medical University
Additional Authors:

- Laura Robinson, MPH, University of Rochester School of Medicine
- Nienke Dosa, MD, MPH, Upstate Medical University
- Adam Dziorny, PhD, University of Rochester School of Medicine
- Kathryn Nobis, BS, University of Rochester School of Medicine
- Michael Nobis, MD, University of Rochester School of Medicine
- Philip Davidson, PhD, University of Rochester School of Medicine

Background: Spina bifida is a complex developmental disability with physical and intellectual impairments that has major impact on a person's well-being, activity and participation. The goal of this study was to evaluate the impact of spina bifida on health status and access to care in a cohort of youth and adults.
Methods: 122 individuals with spina bifida aged 16 to 59 who were seen between January 2007 and December 2008 at the Spina Bifida Center of Central New York were surveyed.

Results: Only 25% of the sample lived independently and 34% were employed. Individuals surveyed had almost seven different affected organ systems. Only 59% reported having good or excellent health; those with fewer organ systems involved had better overall health. They averaged four visits to a specialty health care provider in the 24 month period preceding the study and 71% reported having difficulty accessing health care, which was associated with worse overall health. Hospital admissions and acute visits to the primary care physician were associated with having shunted hydrocephalus, while visits to the emergency department were associated with having a decubitus ulcer.

Conclusion: People with spina bifida are medically complex; they have multiple organ systems involved and report lower than typical overall health. They receive care from multiple providers, and note difficulty accessing services. In the future, people with spina bifida will require access to more medical services, supports and preventive services if they are to have optimal health, well-being and functioning.

Adult Care/Transition Abstracts

4.Title: ***From Transition Challenges to Successes: Establishing a Spina Bifida Adult Care Clinic***
Presenting Author: Betsy Hopson, BA, Children's Hopsital of Alabama
Additional Authors:

- Chevis Shannon-Waters, MBA, MPH, DrPH, Children's Hospital of Alabama, University of Alabama - Birmingham
- Richard Drew Davis, MD, Children's Hospital of Alabama, University of Alabama - Birmingham
- Jeffrey Blount, MD, Children's Hospital of Alabama, University of Alabama - Birmingham
- Patrick Pritchard, MD, University of Alabama - Birmingham

Background: We previously conducted a study to evaluate how patients were transitioned from pediatric and adolescent care to adult care. Our findings were presented at the First World Congress on Spina Bifida meeting. The purpose of our current study was to describe the challenges previously identified and to evaluate our success in providing a more comprehensive and collaborative adult care clinic.
Methods: In 2010 we established a comprehensive adult care clinic facilitating transitional care for many of our spina bifida patients. Benchmarks were developed based on previously identified challenges including: 1) the need to customize a goal-specific action plan for each individual participant based on his/her capabilities, and 2) the establishment of effective interaction between key service providers to facilitate referrals, develop criteria for transition and follow up visits.
Results: With the establishment of our adult care clinic we have developed a lifetime care model for approximately 60 patients with Spina Bifida using the CDC funded electronic medical record. Full support and commitment of our medical director, including monthly participation in the adult clinic, established an environment for success. Additional neurosurgical resources were identified with both pediatric and adult neurosurgeons working together to ensure effective outpatient care and surgical intervention. The pediatric spina bifida coordinator oversees the adult clinic creating continuity of care, facilitating a care plan utilizing previous medical history and addressing unmet needs. Familiar faces and similar practice patterns allows our patients the opportunity to transition with less anxiety and a better understanding of their future medical care.
Conclusion: Collaborations between the pediatric and adult care providers have created an opportunity for successful transition of our spina bifida patients. We will continue to prospectively follow these patients to further determine challenges and successes including improved quality of life and patient satisfaction.

Adult Care/Transition Abstracts

5.Title: ***Health Risk Behaviors among Adolescents and Young Adults with Spina Bifida in Arkansas: A Population-based Cross-sectional Study***
Presenting Author: Minn Soe, MD, MPH, Centers for Disease Control and Prevention

Additional Authors:

- Mark Swanson, MD, MPH, Centers for Disease Control and Prevention
- Julie Bolen, PhD, MPH, Centers for Disease Control and Prevention
- Judy Thibadeau, RN, MN, Centers for Disease Control and Prevention

Background: Persons with spina bifida [SB] who adopt unhealthy lifestyles could be at increased risk of adverse health outcomes because the presence of SB may magnify this risk. The prevalence of health risk behaviors [HRB] among persons with SB is not well known.
Methods: Data analyzed were from 130 young individuals with SB who participated in a 2005 survey conducted by the Arkansas Spinal Cord Commission. We estimated the prevalence of selected HRB, characterized their age distributions and compared their prevalence with national estimates in the U.S general population of same age.
Results: The mean age of participants was 23.5yr (range: 16-31yr) with 66 female and 64 male. Among respondents, 80% lacked regular exercise, 83% watched television ½½3 hrs/day, 75% did not eat healthy food regularly and 49% self-reported as ½overweight'. Twenty eight percent reported cigarette smoking, 28% reported alcohol use, 15% reported binge drinking and 18% reported ever using a drug illegally. Eleven percent reported not seeing a doctor in the year prior to the survey.
The prevalence of self-reported ½overweight', alcohol drinking, and illicit drug use increased with age ($p<0.05$). Adjusted for control factors, adults in their late 20s were more likely to report ½overweight' and illicit drug use than adolescents ($p<0.05$). There was no gender difference in the distributions of HRB. When compared to the general population, respondents were less likely to use tobacco, alcohol and illicit drugs but they tend to eat less healthy diets, do less exercise and engage in more sedentary leisure activities ($p<0.05$ in all comparisons except smoking[$p=0.08$]).
Conclusion: Young people with SB engage in HRB particularly substance use at a later age, and the profile of engagement in HRB is different from the general population. Increased awareness of HRB among people with SB, caregivers and healthcare providers is necessary to promote healthy behaviors.

6.Title: ***Satisfaction with Transition to Adult Health Care: A Focus Group Experience with Adults and Parents***
Presenting Author: Rebecca Boudos, LCSW, Children's Memorial Hospital
Additional Authors:

- Shubhra Mukherjee, MD, Children's Memorial Hospital/ Rehab Institute of Chicago

Background: Focus groups were conducted to better understand patient and parents' experiences transitioning from pediatric to adult care in order to make recommendations for improvements in the process, clinic materials and coordination of care. We will also discuss current efforts at the pediatric level to prepare for transition and how this fits into the 6 core elements of transition from the AAP

guidelines.
Methods: Three sets of focus groups with adults with spina bifida (n=11) and parents (n=11) were conducted, two in person, one via teleconference approximately 90 min each. Groups conducted by staff not associated with the spina bifida program. Groups recorded and qualitative data analyzed.
Results: Similar themes emerged both from the parents and adults. Preparation from Pediatric Side
- Helpful to have information materials to prepare - More planning, preparation, to understand new system Logistics/Clinic Structure
- Infrequency of adult clinic/cancellations/not all doctors present
- Familiarity of some medical staff from pediatric helpful/transition coordinator
- Less wait time during clinic visits positive Emotional Impact of Transition
- Emotional process of transition, letting go of relationships with pediatric team, losing ½family½

Adults: Themes that were stronger for the adult groups included empowerment being in adult care model and recognizing need to take on more responsibility for care and being treated like an adult. Also very interested in finding ways to better prepare patients for transition via peer mentors, greeters, written materials. Parents: Stronger themes for parents included anxiety and fears of unknown as well as recognizing the inevitability of movement to adult care.
Conclusion: Adults and parents both saw positive and negatives in the transition to adult care. Areas for improvement include refining transition preparation from the pediatric side, including transition policy, updating clinic materials, creating possible ½transfer of care mentors½ to help guide new patients, improve access to the clinic. Positives include more independence with health care management, benefits of adult health care setting, some continuity of staff, and shorter appointments.

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Adult Care/Transition Abstracts

7.Title: ***Secondary Conditions in Adults with Spina Bifida***
Presenting Author: Ronna Linroth, PhD, Gillette Children's Specialty Healthcare
Additional Authors:

- Robert Wagner, MD, Gillette Children's Specialty Healthcare

Background: Spina bifida is a condition requiring lifelong rehabilitation and intervention. Eight years of clinical experience at Gillette Lifetime Specialty Healthcare has demonstrated that pressure ulcers, obesity, and urologic conditions significantly impact the health of adults with spina bifida but there has been no quantification of the secondary conditions like these.
Secondary conditions are found to have a negative impact on an individual's autonomy, functional performance, and level of community participation.
Methods: This study used a survey to collect information about secondary conditions experienced by adults, age 18 and older, with spina bifida. Participants were recruited using a purposeful sampling strategy to ensure multiple generation representation.

Results: The first level of data analysis has been descriptive in nature. Complaints of weakness, fatigue, balance, and environmental factors contribute to difficulties in self care. Most startling is the incidence of pain, burns, and wounds. Almost all with depression also reported problems with sleep. Study participants also rated the impact of these conditions on their daily lives.
Conclusion: Understanding the perceived impact of a secondary condition on a patient's life may be influential in the prioritization for intervention and the success in attaining desired treatment and health management outcomes. Patients who are asked direct, specific questions may be more likely to identify difficulties than when asked a general health-related question. The next step of the analysis will be linear regression to learn more about the relationships of the factors. Further discussion on health care planning across the lifespan can benefit early intervention for the prevention or management of secondary conditions.
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8.Title: ***Transition in Young Adults with Spina Bifida: A Qualitative Study***
Presenting Author: Patricia Braun, Dr, Northern Illinois University
Additional Authors:

- Monique Ridosh, Loyola University
- Gayle Roux, Dr, College of Nursing, Texas Woman's University
- Melissa Bellin, Dr, University of Maryland
- Kathy Sawin, Dr, University of Wisconsin

Background: Adolescents and young adults with spina bifida are an at-risk population because of the complexity of their condition, developmental stage and social challenges. The purpose of this qualitative study was to examine the transition to adulthood in young adults with spina bifida and to explore condition related needs and life skills required during the transition process.
Methods: This qualitative study using narrative inquiry was part of a larger multi-site study of adaptation in young adults with spina bifida. Interviews were completed with 10 participants ranging in age from 18-25 years. The guided interview questions focused on specific dimensions of the transition experience related to the ecological model: self-management, independence and inner strength.
Results: Three themes capturing different dimensions of the young adults' transition experience emerged in the analysis. the themes included: (1) Struggling for independence, (2) Limiting social interactions and experiences with stigma, and (3) Building inner strength.
Conclusion: The qualitative study contributes to a better understanding of the challenges of transition to achieve self-management and social development for young adults with spina bifida. Findings in the life stories highlighted issues that necessitate increased advocacy and interventions from professionals within the health and social system.

9.Title: ***Transition to Adult Health Care: Factors Related to the Perception of Health Competency inYoung Adults with Spina Bifida***

Presenting Author: Kim Whitmore, Mrs., University of Wisconsin - Milwaukee College of Nursing
Additional Authors:

- Kathleen Sawin, Dr., Children's Hospital of Wisconsin and University of Wisconsin-Milwaukee
- William Waring, Medical College of Wisconsin/Froedtert Hospital
- Robert O'Conner, Dr., Children's Hospital of Wisconsin
- Merle Orr, Dr., Children's Hospital of Wisconsin
- Karen Orr, Children's Hospital of Wisconsin
- Tera Bartelt, Children's Hospital of Wisconsin

Background: A Spina Bifida Transition Program in a large Midwestern city was developed with a partnership between pediatric and adult providers. Previous research has demonstrated that contextual and process factors may play an important role in transition outcomes. A parallel research project was conducted to evaluate the program and the experiences of young adults with spina bifida (YASB) during the transition.
Methods: The purpose of this longitudinal descriptive study was to explore contextual and process factors related to perceived health competency of YASB without intellectual disabilities. Data was collected for one year from time of transition to adult health care. Measures collected included the Functional Independence Measure (FIM), Family APGAR, Family Environment Scale (FES), Communication and Problem Solving Self-Efficacy Scale (CPSSES) and Perceived Health Competency Scale (PHCS). The Individual and Family Self-Management Theory guided the regression analysis exploring factors associated with PHCS.
Results: The 30 YASB were predominantly Caucasian, the mean age was 22, and 53% were male. Regression analysis revealed that the level of patient mobility was a statistically significant ($p < .05$) predictor of perceived health competence ($B = .092$, $t = 2.686$, $R2 = .21$) but bladder management was not. When CPSSES data was added to the model, level of mobility was no longer significant but self efficacy was ($B = .015$, $t = 3.006$, $R2 = .41$). No relationship between family satisfaction and the PHCS was noted.
Conclusion: Self-efficacy doubled the amount of variance explained. Although YASB that have lower mobility may have a reduced perception of health competency, higher levels of self efficacy may mediate perceived health competency. Healthcare providers working with YASB transitioning to adulthood need to reinforce the importance of increasing self-efficacy in order to positively influence health competence. Future research should investigate the relationship between perceived health competency and self management.

16.

Epidemiology/Genetics Abstracts

1.Title: ***Impact of Hydrocephalus on Health Service Use and Expenditures Among Children with Spina Bifida Enrolled in Medicaid***

Presenting Author: Cynthia Cassell, PhD, Centers for Disease Control and Prevention

Additional Authors:

- Scott Grosse, PhD, Centers for Disease Control and Prevention
- Cora Peterson, PhD, Centers for Disease Control and Prevention
- Phoebe Thorpe, MD, MPH, Centers for Disease Control and Prevention
- Eleanor Howell, MS, Data Dissemination Unit, SCHS, Div Public Health
- Robert Howell, PhD, NC Birth Defects Monitoring Program, SCHS, DivPublic Health

Background: National data on health care use among children with spina bifida (SB) are limited. Available information on health services use and costs typically does not distinguish between children with SB complicated by hydrocephalus and those with SB alone. We examined differences in health service use and expenditures among publicly-insured children in these two groups.

Methods: Children with SB born between 1995 and 2002 were identified by the North Carolina Birth Defects Monitoring Program and linked to Medicaid enrollment and paid claims to identify those continuously enrolled (i.e., enrolled >11 months/year of life). We calculated total Medicaid expenditures per child for medical, inpatient, outpatient, dental, well-child care, SB who survived infancy, 205 (55%) were enrolled in Medicaid. Of these children, 144 were continuously enrolled in Medicaid. After excluding 12 infants who had a birth defect unrelated to SB, health service use and expenditures were assessed for 132 children with SB, 79 with hydrocephalus and 53 without hydrocephalus. During infancy, mean health care expenditure was $33,135 per child with SB, regardless of the presence of hydrocephalus. Mean expenditure for an infant with SB and hydrocephalus was 2.6 times higher than for an infant with SB without hydrocephalus ($40,502 vs. $15,699). The biggest relative expenditures were for developmental/behavioral services (10.1 times higher for those infants with hydrocephalus than without hydrocephalus [$2,278 vs. $225]) and home health services (3.9 times higher [$1,098 vs. $284]). The mean expenditure per child with SB and hydrocephalus was 7.3 times more than for a child with SB without hydrocephalus aged 1"4 years old ($16,042 vs. $2,184).

Conclusion: Comparisons by SB subtype are important for targeting health care resources.havioral services (mental health), and home health for those children with SB with and without hydrocephalus aged 0-4 years old.

Results: Of 373 children with SB who survived infancy, 205 (55%) were enrolled in Medicaid. Of these children, 144 were continuously enrolled in Medicaid. After excluding 12 infants who had a birth defect unrelated to SB, health service use and expenditures were assessed for 132 children with SB, 79 with hydrocephalus and 53 without hydrocephalus. During infancy, mean health care expenditure was $33,135 per child with SB, regardless of the presence of hydrocephalus. Mean expenditure for an infant with SB and hydrocephalus was 2.6 times higher than for an infant with SB without hydrocephalus ($40,502 vs. $15,699). The biggest relative expenditures were for develop-menttal/behavioral services (10.1 times higher for those infants with hydrocephalus than without hydrocephalus [$2,278 vs. $225]) and home

health services (3.9 times higher [$1,098 vs. $284]). The mean expenditure per child with SB and hydrocephalus was 7.3 times more than for a child with SB without hydrocephalus aged 1"4 years old ($16,042 vs. $2,184).
Conclusion: Comparisons by SB subtype are important for targeting health care resources.

2.Title: *Socio-demographic Characteristics and Outcomes in Spina Bifida Patients*

Presenting Author: Michael Schechter, MD, MPH, Emory University, Centers for Disease Control and Prevention
Additional Authors:

- Kevin Liu, MSPH, Centers for Disease Control and Prevention
- Minn Soe, MD, MPH, McKing Consulting
- Elisabeth Ward, RN, MPH, Carter Consulting
- Judy Thibadeau, RN, MN, Centers for Disease Control and Prevention
- Mark Swanson, MD, MPH, Centers for Disease Control and Prevention

Background: This presentation describes a three site process of beta testing a spina bifida electronic medical record (SB EMR) to document site specific use, and identify obstacles encountered and solutions to address them. The overall goal is to share information with other SB clinical sites to aid in broader implementation.
Methods: Three spina bifida clinics (Children's Hospital of Alabama, Children's Hospital Los Angeles, and Children's Hospital Colorado) received funding from the Spina Bifida Association (SBA) to beta test the SB EMR. Technical assistance and consultation was provided by the National Spina Bifida Program at the Centers for Disease Control and Prevention and the SBA. Each site utilizes a different legacy electronic medical record system and has unique operating characteristics, which add to their value as a test site. Regular meetings took place among the group to identify SB EMR implementation challenges, characterize common goals and desired outcomes, and to share potential solutions. The SB EMR is being evaluated for its reporting capability and ability to aid in research, utility in a clinic setting, ability to link with existing hospital EMRs, and use by nonclinical staff.
Results: Data are currently being collected, and the project is scheduled for completion in July 2012.
Early indications are that the SB EMR can be populated by non clinical staff and utilized by clinicians to aid in clinic tasks and flow and for research activities.
Conclusion: Three SB clinics are currently testing a SB EMR to demonstrate utility in the clinical setting and for research. Recommendations will be made at the completion of the project for improvements in the overall process of implementing and utilizing a diagnosis specific EMR, and feedback will be provided to the software company for improvements.

3.Title: ***Spina Bifida Risk is Conferred by Multiple Polymorphisms in Folate One-Carbon Pathway Genes***

Presenting Author: Dennis A. Gilbert, VitaPath Genetics Inc.

Additional Authors:

- Katherine Lazaruk, VitaPath Genetics Inc.
- Jason Stein, VitaPath Genetics Inc.
- Jill Hardin, VitaPath Genetics Inc.
- John S. Witte, Department of Epidemiology & Biostatistics, University of California, San Francisco
- Gary M. Shaw, Department of Pediatrics, School of Medicine, Stanford University
- Edward J. Lammer, Children's Hospital Oakland Research Institute
- Nicholas J. Marini, Department of Molecular & Cellular Biology, University of California, Berkeley
- Jasper Rine, Department of Molecular & Cellular Biology, University of California, Berkeley

Background: Pre-conception supplementation with folic acid clearly decreases the population risk of having an NTD-affected pregnancy. Genetic analysis between the folate one-carbon pathway genes and NTDs has detected few noteworthy associations, with limited replication success. We undertook a casecontrol study of the association between the most common NTD, spina bifida (SB) and variants across 37 genes in the folate pathway.

Methods: 949 Caucasian mothers who had given live birth to a SB affected child and 1,166 matched controls were analyzed. DNAs were genotyped for single nucleotide polymorphisms (SNPs) in 37 genes in the folate-homocysteine pathway. The potential association between each of these SNPs and SB live birth was investigated. Those SNPs showing an association under various genetic inheritance models were used to develop a polygenic risk score prediction model for SB.

Results: DNA sequencing of 37 folate pathway genes in 241 infants with SB identified 824 SNPs suitable for inclusion in the maternal case-control study. 949 case moms and 1,116 controls were genotyped with the 824 SNPs to define risk alleles and develop the prediction model. We found 58 single nucleotide polymorphisms (SNPs) with significant ($p < 0.05$) associations in mothers and SB-affected offspring, providing compelling evidence of a distributed genetic risk signature centered on the folate-one carbon pathway. From these associations, we have developed a risk prediction model. Risk alleles present in 43 SNPs can identify women at 5-fold risk increased for spina bifida affected pregnancy.

Conclusion: The polygenic risk score prediction model provides an important advance in identifying women who are at an increased risk of having a SB-affected child. These findings are under evaluation in an independent population, but strongly suggest women without history of a spina bifida affected pregnancy can be identified as high risk based on their genetic profile.

4.Title: ***Use of Emergency Department Care by Families with Spina Bifida***
Presenting Author: Tera Bartelt, Children's Hospital of Wisconsin
Additional Authors:

- Jenna D. Olstad, Ms, Children's Hospital of Wisconsin
- Evelen M. Kuhn, Ms, Children's Hospital of Wisconsin

Background: Spina Bifida (SB) is a spinal cord dysfunction that contributes to multiple secondary sequelae that can include infections, neurosurgical, urological, orthopedic and dermatologic complications. The SB program at Children's Hospital of Wisconsin (CHW) serves approximately 250 children. The purpose of this study was to determine why children with SB are using the Emergency Department Trauma Center (EDTC), in an effort to identify opportunities to decrease potential nonemergent visits.
Methods: This study is a retrospective chart review of patients admitted to the EDTC with a diagnosis of Spina Bifida between the dates of 1/1/2009 " 12/31/2010.
Results: A total of 303 visits were made by a 133 patients within this time frame. Demographics include: Male 76 (57%), Ages: 0-2 yrs, 39 (29%); 3-5 yrs 16 (12%); 6-12 yrs 41 (31%); 13-18 yrs, 30 (23%) and 19+, 7 (5%), Race: White 91 (68%), Black 22 (17%), Hispanic 15 (11%) Asian 3 (2%) Other 2 (2%). There were 122 (40%) inpatients or short stay admissions from the EDTC. Categories of chief complaints (CC) were Cardiac, Dermatologic, Falls, Fevers, Gastrointestinal, Genitourinary, Musculoskeletal, Neurologic, Respiratory and Other. Top CC of patients admitted to hospital inpatient units or short stay were: Neurologic 41 (33.6%), Gastrointestinal 20 (16.4%), Respiratory 17 (13.9%), Dermatologic 14 (11.5%) and Genitourinary 10 (8.2%). Top CC of patients discharged from the EDTC were: Fever 37 (20.4%), Neurologic 35(19.3%), Gastrointestinal 29 (16.0%), Musculoskeletal and Genitourinary 18(9.9%) each, and Dermatologic 16 (8.8%).
Conclusion: Approximately 55% of our patient population has utilized the EDTC within a two year time period and 40% were admitted. Could some of the 60% remaining visits have been prevented? This study has given us baseline information that will allow the SB Program Team an opportunity to implement strategies to reduce visits to the EDTC.

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5.Title: ***Weight Gain During Pregnancy and Neural Tube Defects in Offspring***
Presenting Author: Wei Yang, MD, Department of Pediatrics, Stanford University
Additional Authors:

- Suzan L. Carmichael, PhD, Department of Pediatrics, Stanford University
- Sarah Tinker, PhD, Division of Birth Defects and Developmental Disabilities, National Center on

Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention

- Gary M. Shaw, DrPH, Department of Pediatrics, Stanford University

Background: Limited information is available about the association of weight gain during pregnancy with risk of birth defects. Our objective was to examine the association of maternal weight gain with neural tube defects (NTDs) among offspring.
Methods: We used data from the National Birth Defects Prevention Study, a multi-center, populationbased case-control study. Mothers of cases and controls were interviewed on many topics, including weight gain. Analyses included 255 anencephaly and 577 spina bifida cases and 5,587 controls with estimated dates of delivery from 1999-2005 resulting from singleton pregnancies to mothers without pregestational diabetes. After subtracting birth weight, we estimated the associations of total and average weekly pregnancy weight gains (kg) with NTDs, stratified by maternal gestational age (<37 vs. >=37 weeks) and adjusted for relevant covariates, including prepregnancy body mass index and race/ethnicity.
Results: Among deliveries >=37 weeks gestation, total weight gain in the lowest quartile (<=6.7kg) relative to the 25th-75th percentiles (6.7-14.9kg) was associated with an increased risk of anencephaly (adjusted odds ratio [aOR]=2.7; 95% Confidence Interval [CI]=1.5, 4.9), but not spina bifida; average weekly weight gain in the lowest quartile was also associated with anencephaly, but not spina bifida.
Among deliveries <37 weeks gestation, a borderline significant association was observed for spina bifida with weekly weight gain in the lowest quartile (<=0.17kg) versus the 25th-75th percentiles (0.18-0.39kg) (aOR=1.6, 95% CI=1.0, 2.4); no association was observed for anencephaly. We consider these latter results descriptive given that cases tended to be delivered earlier than controls, especially anencephaly.
Conclusion: Lower maternal weight gain during pregnancy among births >=37 weeks gestation was associated with anencephaly, and among births <37 weeks gestation it was associated with spina bifida. Lowered weight gain may be a consequence of carrying an NTD-affected fetus or a marker for an underlying factor in the etiology of NTDs.

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6.Title: ***Spina Bifida Risk is Conferred by Multiple Polymorphisms in Folate One-Carbon Pathway Genes***
Presenting Author: Dennis A. Gilbert, VitaPath Genetics Inc.
Additional Authors:

- Katherine Lazaruk, VitaPath Genetics Inc.
- Jason Stein, VitaPath Genetics Inc.
- Jill Hardin, VitaPath Genetics Inc.
- John S. Witte, Department of Epidemiology & Biostatistics, University of California, San Francisco
- Gary M. Shaw, Department of Pediatrics, School of Medicine, Stanford University

- Edward J. Lammer, Children's Hospital Oakland Research Institute
- Nicholas J. Marini, Department of Molecular & Cellular Biology, University of California, Berkeley
- Jasper Rine, Department of Molecular & Cellular Biology, University of California, Berkeley

Background: Pre-conception supplementation with folic acid clearly decreases the population risk of having an NTD-affected pregnancy. Genetic analysis between the folate one-carbon pathway genes and NTDs has detected few noteworthy associations, with limited replication success. We undertook a casecontrol study of the association between the most common NTD, spina bifida (SB) and variants across 37 genes in the folate pathway.
Methods: 949 Caucasian mothers who had given live birth to a SB affected child and 1,166 matched controls were analyzed. DNAs were genotyped for single nucleotide polymorphisms (SNPs) in 37 genes in the folate-homocysteine pathway. The potential association between each of these SNPs and SB live birth was investigated. Those SNPs showing an association under various genetic inheritance models were used to develop a polygenic risk score prediction model for SB.
Results: DNA sequencing of 37 folate pathway genes in 241 infants with SB identified 824 SNPs suitable for inclusion in the maternal case-control study. 949 case moms and 1,116 controls were genotyped with the 824 SNPs to define risk alleles and develop the prediction model. We found 58 single nucleotide polymorphisms (SNPs) with significant ($p < 0.05$) associations in mothers and SB-affected offspring, providing compelling evidence of a distributed genetic risk signature centered on the folate-one carbon pathway. From these associations, we have developed a risk prediction model. Risk alleles present in 43 SNPs can identify women at 5-fold risk increased for spina bifida affected pregnancy.
Conclusion: The polygenic risk score prediction model provides an important advance in identifying women who are at an increased risk of having a SB-affected child. These findings are under evaluation in an independent population, but strongly suggest women without history of a spina bifida affected pregnancy can be identified as high risk based on their genetic profile.

17.

Second World Congress on Spina Bifida Research and Care 2012 . USA, Las Vegas **Tase A**
Neurosurgery/Neuroscience Abstracts

1.Title: ***Knowledge Levels of Staff, Parents and Adolescents with Spina Bifida Regarding Tethered Cord,Shunt Malfunction and Latex Allergy Symptoms***

Presenting Author: Ann Flanagan, PT, PCS, Shriners Hospital for Children-Chicago
Additional Authors:
- Anne Riordan, BA, Shriners Hospitals for Children-Chicago
- Saha Hassani, MS, Shriners Hospitals for Children-Chicago
- Haluk Altiok, MD, Shriners Hospitals for Children-Chicago

Background: Due to limited research, we sought to determine knowledge levels of hospital staff, parents and adolescent patients with Spina Bifida (SB) regarding tethered cord (TC), shunt malfunction, and latex allergy symptoms.
Methods: Demographic and knowledge surveys were completed by 53 hospital staff (7M/46F). During SB clinic, a convenience sample of 37 parents of children with SB (8M/29 F) and 15 adolescent patients (average 15.3 years, 9M/6F) also completed surveys.
Results: Staff: Clinical staff self-rated overall SB knowledge as at least fair. Incorrect responses to increased urinary tract infections (UTI), spasticity, and progressive foot deformities as possible TC symptoms and memory loss as a shunt malfunction symptom was seen for >35%. Over 70% incorrectly answered fever and sneezing as latex allergy symptoms. Parents: One quarter reported no knowledge of TC. Incorrect responses to increased UTI and progressive foot deformities as possible TC symptoms was seen for >68%. Parents of children with shunts reported >35% incorrect responses on all shunt items. Over 35% of parents did not know that eye irritation and wheezing were latex allergy symptoms or that disposable diapers, wheelchair and crutch parts, and sport equipment handles may contain latex. Patients: Fifty-three percent of patients had no knowledge of TC and answered all TC items incorrectly >60% of the time. Of the 47% of patients with shunts, >50% knew the malfunction symptoms of headache, nausea or vomiting, and redness or swelling along shunt track. However, <50% knew the remaining eight common symptoms. Besides itching, >50% were unaware of any symptoms of latex allergies as well as items that contain latex.
Conclusion: There were marked knowledge gaps for all groups in the areas of TC, shunt malfunction, and latex allergy. The adolescents had the least awareness. Improving education for all groups will be beneficial for the care of patients with SB.

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Neurosurgery/Neuroscience Abstracts

2.Title: ***Selective Ventriculoperitoneal Shunt Placement in Myelomeningocele: If It is Good for theBrain is it Bad for the Bladder?***
Presenting Author: Elizabeth Yerkes, MD, Children's Memorial Hospital
Background: The recent MOMS Trial described lower incidence of ventriculoperitoneal shunt (VPS) after prenatal closure. With a goal of reducing morbidity and mortality associated with VPS, our team has implemented new criteria since 2002 for selective VPS placement after neonatal closure. We evaluate the impact of selective VPS on urologic status.
Methods: All children with neonatal closure of myelomeningocele (MM) at our institution from 2002 to 2010 (Group A) were compared to a contemporary comparison group 2006-2010 (Group B) closed locally but enrolled with our team since early infancy. Group A was subdivided into Shunted and Unshunted subgroups based upon early VPS decision. Baseline radiologic status, cystometrogram profile (CMG) and subsequent changes in urologic or neurologic status were noted. Proactive medical management was utilized as indicated by CMG. At least one year of follow-up and two CMGs were required for inclusion. Logistic regression was performed.
Results: Mean follow-up was 6.3 years in Group A Shunted, 4.9 years in Group A Unshunted, and 3.3 years in Group B. VPS was inserted in the neonatal period in 54% of Group A and 100% of Group B

(Chi-test, p=0.009). Revision was required in 76% of Group A Shunted (range 1-18 revisions, mean 4.1) and 73% of Group B (range 1-6 revisions, mean 2.5). Mean time to radiologic or CMG deterioration was 1.2 years, 1.6 years and 1.4 years in Groups A Shunted, Group A Unshunted and Group B, respectively. No statistically significant differences were found between the subgroups of Group A and Group B.
Conclusion: VPS malfunction is a common source of morbidity in MM. Close early childhood urologic follow-up does not reveal any apparent detriment to the urinary tract with selective VPS approach. Careful longitudinal follow-up of neurodevelopmental and urologic progress is required to validate this approach.

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Neurosurgery/Neuroscience Abstracts

4.Title: ***Transitional Care in Neurosurgery: Follow up from a 12 Month Experience***
Presenting Author: Jeffrey Blount, MD, UAB/Childrens of Alabama
Additional Authors:

- Betsy Hopson, Childrens Hospital of Alabama
- Chevis N Shannon, MBA, MPH, DrPH, UAB
- Kristen Riley, MD, UAB
- Mamehri Okor, MD, UAB
- Patick Okor, MD, UAB

Background: Transition of care from pediatric to adult care is an important and timely challenge in Spina Bifida (SB). We recognized that the needs of patients who were transitioning were being insufficiently met. We therefore developed a paradigm for pediatric neurosurgical coverage in an already existing adult spina bifida clinic and report here our initial twelve months experience.
Methods: Our previous review of the transition process confirmed that we were not successfully transitioning patients to adult care. Successful transition was defined as attendance of follow up visits, appropriate interaction with adult providers, and patient satisfaction.
Results: In October of 2010 we established the Adult Spina Bifida Clinic at UAB. The medical director of the pediatric spina bifida clinic agreed to participate in the adult clinic allowing for better continuity of neurosurgical care and institutional memory that enhanced the adult neurosurgeons ability to facilitate appropriate neurosurgical intervention. Since establishing the adult care clinic 60 adult patients have been seen, reviewed and evaluated. Four adult patients required further evaluation for signs of tethered spinal cord (TS only one of these went on to require shunt revision. Patient satisfaction with the transition experience was improved and transition was markedly facilitated. Resistance to depart the pediatric environment dissipated with assurance of ongoing competent care.
Conclusion: Multiple models of successful transition exist. Neurosurgery needs are often most challenging because of the potential acuity and severity of shunt problems and a paucity of adult providers experienced in the nuances of care for patients with SB. The described model has worked well but will be contrasted with other described paradigms from other centers. Acute need for care appears reduced in the adult population followed to date.

5.Title: ***A World of Opportunity: Disparities in Spina Bifida Care and Neurosurgical Evidence-Based Global Health Practice***
Presenting Author: Jonathan Castillo, MD, MPH, Cincinnati Children's Hospital Medical Center
Additional Author:

- Besma Abbaoui, PhD, University of Cincinnati, College of Medicine
- Heidi Castillo, MD, Cincinnati Children's Hospital Medical Center

Background: New technologies continually advance the standard of care for patients with Spina Bifida (SB). Recently prenatal interventions for myelomeningocele have received considerable attention among developed nations; nevertheless, less attention has been given to the implementation of more traditional approaches to hydrocephalus and comorbidities' management in low-resource settings. The purpose of this study was to explore the history and current forms of neurosurgical management guidelines in SB care across the globe.
Methods: A PubMed search was performed for articles on neurosurgical management in patients with SB. Articles were identified by searching for the terms "spina bifida" and "neurosurgery." Articles were excluded if they did not provide practice recommendations; the included publications were further focused on review articles and meta-analyses. Articles meeting inclusion criteria were classified according to management focus and the country of origin. The authoring countries were stratified by World-Bank specified income level.
Results: A total of 958 articles were identified, of which, 79 met inclusion criteria. From 1975 until 2011, there was observable growth in the number of publications on neurosurgical management; the range of number of publication per year was zero to twelve. Institutions in nations with high, upper-middle, and lower-middle income economies published 89.9%, 3.8%, and 6.3% of the guidelines respectively. Authors from countries represented outside of high income economies included Argentina, China, India, Nigeria, Taiwan, and Uganda.
Conclusion: In light of numerous technological advances in pre-natal and neo-natal neurosurgical care, few clinical practice guidelines in the management of SB patients have been written outside high-income countries. Thus there is lack of care guidance which is culturally and technologically appropriate for 85.4% of the world's population. This represents an opportunity for academicians in high-income countries to more closely collaborate with their counterparts in middle and low-income nations in order to gain from their perspective and co-author guidelines which are technologically appropriate for a vast portion of the globe.

7.Title: ***Complications Following Tethered Cord Surgery in Spina Bifida Patients***
Presenting Author: Virginia Briggs, PhD, University of Massachusetts Medical School
Additional Author:

- Oguz Cataltepe, MD, University of Massachusetts Medical School

- Patricia Franklin, MD, University of Massachusetts Medical School

Background: Tethered cord syndrome (TCS) is a functional disorder of the spinal cord that commonly occurs in people with spina bifida. Surgery to treat TCS has been reported to occur in 13% to 25% of spina bifida patients. Complications resulting from TCS surgery have been documented in the literature, but not specifically in the SB population. The goal of this study was to quantify post-operative complications following TCS surgery in the SB population, and explore patient characteristics using a large national hospital discharge database (Nationwide Inpatient Sample) over a ten year period (1999-2008).
Methods: The Nationwide Inpatient Sample (NIS), the largest inpatient database publicly available in the United States and containing 20% of all hospital stays was used. Data was extracted from 1999 to 2008 and merged, and ICD-9 diagnosis and procedure codes were used to identify patients. Outcome measures included complications, days in hospital, hospital discharge status diagnoses and procedures.
Results: Of 44,719 patients diagnosed with spina bifida, a total of 1,420 patients who underwent surgery for TCS were identified. The mean age was 26.9 years, 56% were female and the mean hospital stay was 6.8 days. Post-surgical complications were significantly higher in older patients (>21 years). Number of neurologic complications increased with age, and was highest in patients 30-40 years (5%). Neurologic complications, neurogenic bladder, urinary tract infection, hypertension, number of diagnoses and procedures were significantly higher in patients with higher level lesions.
Conclusion: Increased age and severity of spina bifida (thoracic to cervical level lesions) were associated with increased post-surgical complications and comorbidities in patients who underwent surgery for tethered cord. Identifying patients at higher risk for post-surgical complications will allow physicians to alter treatment plans to lower these risks before performing surgery.

18.

Orthopedics Abstracts

1.Title: ***Correlation Between Ventriculoperitoneal Shunt Complications and Functional Gait Quality in Pediatric Patients with Myelomeningocele***
Presenting Author: Vineeta Swaroop, MD, Northwestern University Feinberg School of Medicine, Department of Orthopaedic Surgery
Additional Authors:

- Eric Szczesniak, BS, Children's Memorial Hospital
- Melissa Colthrust, KT, Children's Memorial Hospital
- Laura Johnson, MS, Children's Memorial Hospital
- Claudia Kelp-Lenane, PT, Children's Memorial Hospital
- Luciano Kelp-Lenane, MD, Northwestern University Feinberg School of Medicine, Department of Orthopaedic Surgery

Background: Up to 90% of children with myelomeningocele (MM) and concomitant hydrocephalus require placement of a ventriculoperitoneal shunt (VPS). Recent data have shown children with MM and no shunt walk at a significantly greater velocity and stride length as compared to those with a shunt. However, the effect of VPS complications, specifically revision and infection, on gait has not been previously described. Our goal was to test 2 hypotheses: 1- an increased frequency of VPS revisions leads to decrease in functional gait parameters; 2- an incidence of VPS infection leads to decrease in functional gait parameters compared to no shunt infection.
Method: Patient records were reviewed from 1987 to 2005, and 96 patients age 4 to 18 years met inclusion criteria. Excluded were age <4 years at time of assessment, no VPS present, and VPS complication within 6 months of evaluation. Data collected included Functional Mobility Scale (FMS), lesion level, and number and dates of 1) VPS revisions and 2) VPS infections. For a subset of 61 participants who underwent a three-dimensional gait analysis, we also collected temporal-spatial gait parameters (velocity, cadence, and stride length). Regression analysis was used to correlate VPS revisions or infections with FMS and gait parameters and adjusted for confounding by age, gender, lesion level, orthotic use, and presence of scoliosis.
Results: When VPS revision was performed after the age of 2 years, no significant differences were noted in FMS or gait parameters based on number of shunt revisions. However, the 11 with VPS infections showed a negative trend in gait parameters and a statistically significant decrease in FMS 50 ($p=0.029$) and FMS 500 ($p=0.003$) compared to patients without infection.
Conclusion: In the absence of a VPS infection, the FMS and gait parameters are not affected by number of shunt revisions. However, VPS infection has a negative effect on FMS and gait parameters.
Specifically, patients with VPS infection were less independent at distances representing mobility in school and in the community compared to patients without infection. This information allows caregivers to counsel patients with MM and VPS regarding functional ambulatory expectations.

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Orthopedics Abstracts

2.Title: *Longitudinal Temporal-spatial Gait Assessments of Myelomeningocele Patients: Does BMIAffect Long-term Functional Status?*

Presenting Author: Vineeta Swaroop, MD, Northwestern University Feinberg School of Medicine, Department of Orthopaedic Surgery
Additional Authors:

- Christine Cherella, BA, Children's Memorial Hospital
- Tasos Karakostas, MPT, PhD, Children's Memorial Hospital
- Luciano Dias, MD, Northwestern University Feinberg School of Medicine, Department of Orthopaedic Surgery

Background: Research has shown myelomeningocele (MM) patients can have higher percentages of body fat, but data correlating weight to function over time is lacking. Our goal was to investigate the

long-term ambulatory prognosis of MM patients in terms of gait parameters; and to assess if body mass index (BMI) differences affect these parameters.
Method: A retrospective chart-review was conducted to identify MM patients undergoing gait analysis since 1989. Included were patients undergoing at least 2 studies at least 5 years apart. Temporal-spatial gait parameter data (velocity, cadence, and stride length) was obtained and expressed as percentages of established age-matched norms. BMIs were calculated using data from each visit. Excluded were patients with: less than 5 years follow-up, additional diagnoses (tethered cord, shunt infection), initial study at <5 years of age.
Results: 21 patients were reviewed, 12 sacral-level and 9 low-lumbar level. Average initial and final age: sacral patients was 9+2yr and 16+1yr, low-lumbar patients was 8+2yr and 15+1yr. 75% of sacral patients showed an overall increase in velocity, 67% showed increase in cadence, and 83% showed increase or maintenance in stride length. Sacral patients had an average initial and final BMI 69.5% and 64.9%. 44% of low-lumbar patients showed an overall increase in velocity (56% showed a decrease), 44% showed increase in cadence, and 89% showed increase or maintenance of stride length. Low-lumbar level patients had an average initial and final BMI of 86.9% and 81.2%.
Conclusion: Sacral patients have potential to maintain or increase velocity, cadence, and stride length over time while low-lumbar patients are less predictable. Some showed improvement in velocity or cadence, whereas others did not which may be related to energy cost issues as low-lumbar patients overall had higher BMIs. With this knowledge, we have the ability to counsel patients and families regarding ambulatory prognosis. In addition, noting the BMI differences, we can provide guidance at an early age, especially to low-lumbar patients, to help avoid obesity.

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Orthopedics Abstracts

3. Title: *Spinal Deformity Correction in Conjunction with Tethered Cord Release*

Presenting Author: Haluk Altiok, MD, Shriners Hospitals for Children-Chicago
Additional Authors:

- Anne Riordan, BA, Shriners Hospitals for Children-Chicago
- Adam Graf, MS, Shriners Hospitals for Children-Chicago
- Joe Krzak, PT, Shriners Hospitals for Children-Chicago
- Sahar Hassani, MS, Shriners Hospitals for Children-Chicago

Background: Spinal cord tethering that may occur after surgical repair of myelodysplasia is a common cause of rapid progression of spine deformity. Release of tethered cord may address further progression of spine deformity without additional procedures. This study was designed to describe the progression of spine deformity in this population with and without surgical intervention following tethered cord release.
Methods: Medical charts of 96 patients who underwent tethered cord release between 1980 and 2010 were reviewed. Of the 35 patients that presented with a chief complaint of progressive spine deformity, 18 underwent further corrective spine surgery (avg. age 9.4 +/- 3.5 yrs.) after the tethered cord was released while 17 were managed conservatively (avg. age 5.7 +/- 3.9 yrs.) after the tethered cord was released. Cobb angles were measured at follow-up clinic visit after corrective spine surgery for Corrective Spine Surgical Group (0.7 +/- 0.9 years) and after conservative treatment for Non-corrective Spine Surgical Group (1.2 +/- 1.1 years).

Results: Average Cobb angle of the Corrective Spine Surgical Group was 68.4 +/- 21.8 degrees before the tethered cord was released and 44.5 +/- 16.6 degrees after corrective spine surgery. Average time between tethered cord release and corrective spine surgery was 0.7 +/- 0.7 years; average time between tethered cord release and follow-up Cobb angle measure was 1.4 +/- 1.4 years. Average Cobb angle of the Non-corrective Spine Surgical Group was 41.6 +/- 15.1 degrees before the tethered cord was released and 45.6 +/- 20.8 degrees after the tethered cord was released. In the Non-corrective Spine Surgical Group, scoliosis of 2 (11.8%) patients improved, 12 (70.6%) remained the same, and 3 (17.6%) worsened.
Conclusion: The results demonstrated that tethered cord release without additional corrective spine surgery may be able to adequately stabilize spinal deformity in patients with myelodysplasia.

19. Title " Schlechte Nachrichten nach vorgeburtlicher Untersuchung" **Tase D**

Eine Begleitschrift für Frauen und Paare, die einen Schwangerschaftsabbruch in Erwägung ziehen.
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20. „ Interdisziplinäre Behandlung bei Spina Bifida notwendige Versorgungsstrukturen"

Saksamaa Berliini Charite Ülikooli Myelodüsplaasiahaigete Keskuse struktuur ja põhimõte: (tõlkes. Spina Bifida haige kui interdistsiplinaarse haige raviks vajalik struktuur ja ravimiseks korralduslik erinevate instantside ülesehitus)

Einleitung: Kinder, Jugendliche und Erwachsene mit Spina bifida sind für eine umfassende Behandlung und Unterstützung auf eine Vielzahl von verschiedenen Fachleuten und Institutionen angewiesen – allein das Inhaltsverzeichnis dieser Broschüre gibt einen Eindruck vom Umfang und von der Vielgestaltigkeit der Probleme. Ein tragfähiges, für den Einzelnen passendes Behandlungskonzpekt kann jedoch nur entstehen und in die Tat umgesetzt werden, wenn eine koordinierte Zusammenarbeit von erfahrenen Spezialisten gewährleistet ist. Der folgende Beitrag beschäftigt sich mit den Grundlagen, die für Interdisziplinäre Zusammen-arbeit besonders wichtig erscheinen , und den Strukturen , die zur Umsetzung benötigt werden.
Grundlagen der interdisziplinären Versorgung
1.Zielsetzung. Die Interdisziplinäre Behandlung hat zum Ziel, für jeden Betroffenen grösstmögliche Autonomie und möglichst umfassende Teilhabe arm gesellschaftlichen Leben zu erreichen.

21.

" The Spina Bifida network in France" **Tase D**

Presenting Author: Manunta A, Jezequel M, Hubert J, Menard H Centre de Référence Spina Bifida

How manyspina bifida patients in France?
2010 –2014 : 6500 spina bifida patients have been hospitalized at least once
Everyyear100 –130 newbornspina bifida patients

French national plan for rare diseases
«Assurer l'équité pour l'accès au diagnostic, au traitement et à la prise en charge»
2005-2008 : 1st national plan
Budget: 100 millions €
- **Improvementof medical knowledge and management**
- **Epidemiology (national database on rare diseases)**
- **Prevention**
- **Information (Internet site «Orphanet»)**
- **Developand coordinateaction at european level(research programs, European guidelines...)**

Key role of patients associations
FEDERATION FRANÇAISE DES ASSOCIATIONS POUR LE SPINA BIFIDA

Concentrate management of rares diseases on few centers of excellence

Improve medical expertise

Improving clinical practical guidelines

Develop and coordinate research

Inform
- Health professionals
- Patients
- Public at large

National center of expertise in spina bifida in Rennes
The «Centre de Référence» Spina Bifida in Rennes:
Labelledin 2007, Recertifiedin 2012 **Budget: 210.000 €/year**
A network of «centres de compétence» at régional level 9 centers for spina bifida
No budget

Multidisciplinary meeting
Conclusion of the evaluation
Conclusion mailed to the patient, his GP and specialists
Personalised care program sent to the patient
login + password sent to patient and GP to give access to electronicmedical records
The patient is referred to the regional «centre de competence» nearest to where he lives
Centre de référence
RECOURS

Conclusion
The French spina bifida network within the French rare disease plan has:
- improved management of spinal dysraphism(multidisciplinary clinics, transition clinics childhood→adulthood…)
- Improved accessto care within a nationlnetwork for patients
- Increased awareness of the pathology
- Impulse the researchon spinal dysraphism

At a reasonable cost for social security

21.a Eesti keelne kordus:

PRANTSLASTE ETTEKANNE if-i WEB seminarist 19.01.2016
versus EESTI MDPK samal seminaril seisul 19.01.2016.

Prantsusmaa alustas

RAHVUSLIKu PLAANi koostamisega HARVAESINEVATE HAIGUSTE suhtes

Aastatel 2005 – 2008 oli eelarveks 100 MILJONIT EUROT
- EPIDEMIOLOOGIA

(rahvusliku harvikhaiguste registri loomiseks)
- INFORMATSIOONILE (interneti lk.)
- ARENDUSE JA KOORDINEERIVAD TEGEVUSED EUROOPA TASANDIL
(teadusprogramm uuringutele, euroopalike juhiste väljatöötamisele)
- KONTAKTIDE ÜLESVÕTT PATSIENTIDE ESINDUSORGANISATSIOONIDEGA

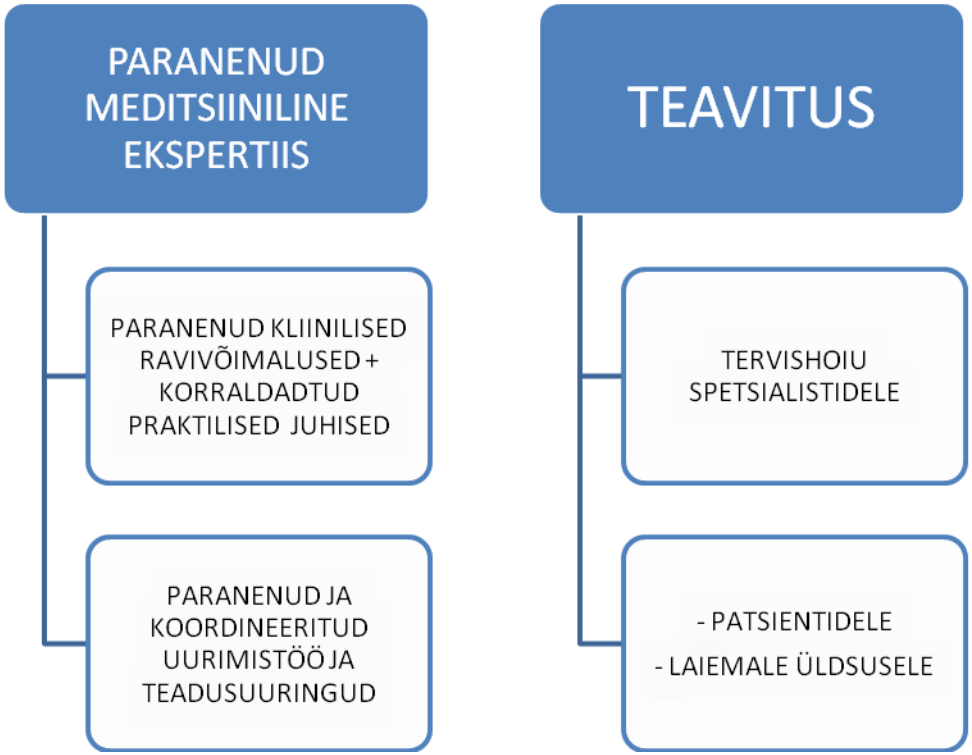
PRANTSUSMAA RAHVUSLIK PLAAN HARVAESINEVATE HAIGUSTE OSAS
world factboock 2016.a.

o Eesti 45 399 pindala km2	- Prantsusmaa 547 000 km2 (lisaks on veel meretagused alad)
o Elanike arv ~ 1.3 miljonit	- Elanike arv ~ 62 miljonit
o Iseseisvus 24.02.1918. ja taasiseseisvus 20.08.1990.	- Iseseisvus 06.06.1523.
o Eesti Seljaajusonga ja Vesi-peahaigete Selts 03.01.1991.	- Renne`s SpBH keskus 2007.a., milline rekonstrueeriti 2012.a.
o Eestis SKT sisekogutoodangu produkt 2,916 miljardit USD	- Prantsusmaa SKT on 2,108 triljonit USD
o Ühe elaniku kohta SKT	- Ühe elaniku kohta SKT USD-s
EESTI 29 500 US dollar	PRANTSUSMAA 33 678 US dollar
LÄTI 25 700 “	USA 57 300 “
LEEDU 29 900 “	LUXEMBOURG 102 000 “

PRANTSUSMAA RAHVUSLIK PLAAN HARVIKHAIGUSTELE
kontsentreerida harvikhaiguste alane juhtimine üksikutesse headesse keskustesse

Spina bifida referentskeskus Rennes`s
- Loodud 2007. a.
- Restruktureeritud 2012.a.
- Aasta eelarve 210 000 eurot
- Koondab enda juurde 9 väiksemat lokaalset SpB keskust üle Prantsusmaa, kelledel ei ole omaette iseseisvat eelarvet ja nõustab ka teisi ligilähedase patoloogia keskusi

REFERENTSKESKUSE STRUKTUUR JA ÜLESEHITUS



Renne´e SpBH Keskus

- Arvel 2015. a. **371 haiget**
- Alustasid 12 haigest ja arvukus on lineaarselt suurenenud
- Vanuseliselt on suurim osakaal 18-24 a.vanusel (noorematel sündivus väiksem ja vanemad juba surnud paljud)
- KOHALIKUL e. REGIONAALSEL TASEMEL
- Keskus korraldab oma regiooni alakeskustele igal kuul videokonverentsi
- Haigusjuhtude arutelu
- Patsientide konsultatiivne nõustamine

- Töötavad RAHVUSLIKUL ja ÜLEPRANTSUSMAALISEL TASEMEL
- Koordineeriv koostöö teiste harvikhaiguste referentskeskustega:
- Hõlmavad ka teised ligilähedased NTD-ga seotud harvikpatoloogiad
  . syringomyelia
  .kongenitaalsed perineal prk-a malformatsioonid
  .anorektaalsed väärarengud

22.

TEEMA: Spina bifida

Materjali koostas: lastekirurg Ann Paal *Eesti Puuetega Inimeste Koja tarvis iseloomustamiseks EPIK`u allorganisatsiooni Eesti Seljaajusonga ja Vesipeahaigete Seltsi*

1. Spina bifida – definitsioon, avaldumine ja tekkepõhjused

Spina bifida e. lõhestunud selg (kreeka keelse mõiste otsetõlge) märgib olukorda, kus kaasasündinud arengurikkest on lülisamba üks või mitu lülikaart jäänud normaalselt sulgumata. Esimesel raseduskuul moodustub inimlootel tulevane kesknärvisüsteem e. pea- ja seljaaju, mis normaalsel juhul kattub 24. raseduspäevaks ringjalt luulise ümbrisega. Peaaju ümbritsevad koljuluud ja seljaaju lülisambalülid. Vahel häirub normaalne „luulise ümbrise“ moodustumine kas üksikute luude anormsuse või luude omavahelise kokkupuute vea tõttu nii, et luukate jääb „lõhestunuks“. Selliselt defektselt alalt võib eenduda songataolise sopistusena pea piirkonnas peaaju ja selja piirkonnas seljaaju. Pea osas räägime peaajusongast ja selja piirkonnas seljaajusongast. Reeglina on väärarengulise piirkonna kõik koed sh ajukude ise ka teatud moel normist erinevad. Ühtset täpset kesknärvisüsteemi arenguanomaalia tekkepõhjust tänapäeval veel ei teata. Peamiseks põhjuseks loetakse vitamiin B9 e. foolhappe defitsiiti raseda organismis. Inimloote arengus on vitamiin B9 molekulid rakkude paljunemise, diferentseerumise ja nendest elundsüsteemide moodustumisprotsesside käivitajaks ning kontrollijaks. See arengurike on seotud elukestvate mitmesuguste halvatusprobleemidega, millistest tervistuda ei saa, aga on võimalik kaasaegse ravi ning abivahendite abil leevendada.

2. Puude esinemissagedus

Spina bifida loetakse harvaesinevaks haiguseks, sest enamus närvisüsteemi varase etapi väärarenguga seotud loodetest hukkub. Euroopas sünnib igal aastal ~4500 last selle arenguanomaaliaga. Eestis ~ 4-6 last Spina bifida haiget aastas.

3. Kaasnevad erivajadused

Pea- või seljaaju väärarenguline piirkond on lisaks anatoomilisele iseärale muutunud loomu tõttu vahetult seotud mitmete organsüsteemide normist erineva talitlusega. Väärarengulist ala läbivad närviimpulsid takistuvad siin ja kõik kahjustusalast

allpoole jäävad piirkonnad on toimimiseks vajaliku normaalse infota. Reeglina avaldub see mitmesuguste halvatus-nähtudena: lihastoonuse muutusest tekivad jäsemete asendideformatsioonid, käimisvõimetus, põie- ja sooletöö talitushäire uriini või roojapidamatusena. 80 % haigetest esineb ka vesipea ehk hüdrotseefalus ehk „ajuvedelik ei mahu pähe ära“ nagu üks noormees selgitas teisele. Vesipea raviks kannab enamus kehasiseseid shundisüsteeme. Shundid on kunstlikud torud, mis juhivad liigse ajuvedeliku peast keha teise piirkonda. Shundisüsteem seab haigele mitme-suguseid režiimipiiranguid. Sageli esineb nägemishäireid. Kognitiivses plaanis on spina bifida patoloogiaga inimestel ealise normi keskmisest enam valikulist üliandekust või alaandekust. Kaasnevateks puueteks võivad olla kõnetakistused, psühholoogilised probleemid, suhtlemis-hirmud, ärevushäired, tähelepanu koondamise raskus, lineaarset tüüpi õpivõimekus st. mitme-le asjale korraga ei suuda tähelepanu jaotada, epilepsia e. kramptõbi, tavanormist kiirem väsi- muse teke nii vaimsel pingutusel, aga ka füüsilisel pingutusel, eakohaselt ise oma hooldus-probleemidega mitte hakkamasaamine, lamatised ja sagedased põletikulised haigused, piira-tud liikumisvõime. Haiged / õigem on öelda spina bifida tõttu erivajadustega inimesed/ vajavad elukestvalt multiprofiilset eriarstiabi ja rehabilitatsiooniteenuseid ning puudelisust kompenseerivaid erinevaid abivahendeid ilma tavajärjekorras ootamiseta, et püsida võima-likult heas tervisseisus ja töövõimelisuses. Kuigi spina bifida multiprofiilsed haiged on meditsiinile ja ka sotsiaalsüsteemile „väga kallihinnalised“ otseses rahalises mõttes, on puu-dest tulenevate tüsistuste korral hind 10x kallim.

4. Takistused tööturul osalemisel

Ei saa tavamõistes füüsilist tööd teha nn nõrga selja tõttu. Töökoht peaks olema individuaalse sobitusega. Sageli vajavad haiged teistest enam juhendamist ning tööesi ettevalmistamist. Olenevalt võimekusest tuleb proovida leida individuaalsele võimekusele vastav sobiv töö, töötamise vorm ja kohaldatud töökoht. Hea on töö, kus ei ole normi ja kiirustamist, on privaatsuse võimalus. Põie- ja sooletöö häire esinemise juures ei sobi toidu käitlemise ja toidu valmistamisega seotud töökohad. Epilepsia korral ei sobi töö veekogu vahetus läheduses, liiklusvahendi juhtimine, liinitöö kiirelt liikuvate objektidega või mehhanismidega. Olenevalt kuulmislangusest või sõnaoskusest ei sobi töö klienditeeninduse valdkonnas. Kõrgustes töö-tamine võib raske olla tasakaaluhäirete ja ruumitajuhäirete tõttu. Ortoosides või ratastoolis töötaja peaks arvestama juurdepääsuvõimalustega, treppidel liikumise oludega, kohandatud töökoha vajadusega ja töö juurde korraldatud puhkamisvõimalustega. Üldjuhul on erivajadusega tööealiseks sirgunud ja töövõimelised haiged töösituatsioonis ettevaatlikumad ja truumad oma tööle kui teised. Neid on väiksest peale õpetatud oma puudelisuse, keelatud või lubatud piirangute juures toimetulekuga arvestama.

5. Takistused igapäevaelus

Igapäevaelus hakkamasaamine sõltub puude sügavusastmest ja hulgiprobleemide hulgast: liikumisprobleemid (ratastooli vajadus, ortoosid, transport, juurdepääs), suhtlemisvõimekus (raskuste korral ei sobi näiteks suures kollektiivis töö jt), iseenesega hakkamasaamise võimekus (sh soole- ja põietöö üle kontroll), vaimne tase, motoorika, kõrvalabi vajadusel selle puudumine.

6. Vajalikud abivahendid ja teenused töö- ning igapäevaelus.

Spina bifida patoloogia puudelisuse aste võib olla kergest vormist väga sügavani. Sellest sõltuvalt on ka abivahendite vajadus väga laiaskaalaline. Väga olulised on kontinentsi hoidmiseks abivahendid ja tingimuste korraldus, liikumisabivahendid sh. ortoosid,

ratastool, transport, vaegnägemisel prillid, vahel kuuldeaparaat, arvutid ja automatiseeritus, mobiil-telefon ja internetiseeritus, erilahendused häiresüsteemidele nagu signaallambid, vibreerivad kellad, madalama helisagedusega suitsuandur; kommunikaatorid, motoorset kohmakust kom-penseerivad vahendid, puudelisuse kompenseerimisena kohandatud kodusisustus, kohandatud töökoht sh samuti sobilik tualettruum.

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