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Author: Bergen, Janneke van den

Title: Diversity in disease course of Duchenne and Becker muscular dystrophy

Issue Date: 2015-06-24



A Appendices

List of abbreviations

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List of abbreviations

6MWT	Six minute walk test
AON	Antisense oligonucleotide
ASAT	Aspartate aminotransferase
ALAT	Alanine transaminase
BMD	Becker muscular dystrophy
cGMP	Cyclic guanosine monophosphate
DCG	Dystrophin associated glycoprotein complex
DCM	Dilated cardiomyopathy
DDD	Dutch Dystrophinopathy Database
DMD	Duchenne muscular dystrophy
DNA	Deoxyribonucleic acid
ECG	Electrocardiogram
EMA	European medicines agency
FS	Fractional shortening
FVC	Forced vital capacity
LOVD	Leiden Open Variation Database
LTBP4	Latent transforming growth factor 4
LVEDD	Left ventricle end systolic diameter
LVESD	Left ventricle end systolic diameter
LVF	Left ventricular function
MRI	Magnetic resonance imaging
mRNA	Messenger ribonucleic acid
MVIC	Maximal voluntary isometric contraction
NA	Not available
NMD	Neuromuscular diseases
nNOS	Neuronal nitric oxide synthase
PSA	Prostate specific antigen
QMT	Quantitative muscle testing
ROI	Region of interest
SD	Standard deviation
SNP	Single nucleotide polymorphism
SPP1	Secreted phosphoprotein 1
TA	Anterior tibial
TGF	Transforming growth factor
UK	United Kingdom

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Curriculum vitae

Janneke Carolina van den Bergen werd op 14 oktober 1979 geboren in Leiden. Ze behaalde in 1997 cum laude haar VWO-eindexamen aan de Heertganck te Heerde. In 1999 startte ze met haar studie geneeskunde aan de Rijksuniversiteit Groningen. Tijdens haar studie was ze werkzaam bij de Kinderleefbaarheidsdienst. Daarnaast was ze actief bij de International Federation of Medical Students' Association (IFMSA). Na diverse lokale functies werd ze in 2002 verkozen tot voorzitter van het nationaal bestuur. Na haar aftreden in 2003 nam zij tot 2005 zitting in de Raad van Toezicht. Als liaison officer voor UNICEF was zij in 2003-2004 tevens verbonden aan het internationale bestuur van de IFMSA.

In het kader van haar wetenschappelijke stage verbleef Janneke in 2003 een half jaar in Washington DC, waar ze in het Children's National Medical Center onderzoek verrichtte naar de rol van de IGF-bindingseiwitten in spierdystrofie (begeleiding: prof. dr. EP Hoffman). Na haar co-schappen in Zwolle behaalde ze in 2005 cum laude haar arts-examen. Aansluitend begon ze in januari 2006 haar opleiding tot neuroloog aan het Leids Universitair Medisch Centrum (opleiders: prof. dr. RAC Roos en prof. dr. JJ van Hilten). In november 2007 startte ze onder leiding van prof. dr. JJGM Verschuuren haar promotie onderzoek naar Duchenne en Becker spierdystrofie, zoals beschreven in dit proefschrift. In 2012 werd haar poster over de relatie tussen ziekte-ernst en dystrofine in Becker spierdystrofie tijdens het congres van de World Muscle Society in Perth, Australië uitgekozen voor een plenaire presentatie. Om zich verder te verdiepen in de kinderneurologie was Janneke in 2014 gedurende een half jaar werkzaam in het Sophia kindziekenhuis te Rotterdam (begeleiding: dr. CE Catsman). Sinds 2013 is ze betrokken bij het ontwikkelen van medisch studiemateriaal voor diverse opleidingen van het NTI. Ze hoopt haar opleiding tot neuroloog in september 2015 af te ronden.

Janneke woont in Oegstgeest, samen met haar man Robert en hun kinderen Babette, Quirine en Elias.

Dankwoord

Dit proefschrift was niet mogelijk geweest zonder de hulp van velen. In het bijzonder gaat mijn dank uit naar de jongens en mannen met Duchenne en Becker spierdystrofie die belangeloos informatie over hun ziekte hebben verstrekt en deelnamen aan diverse onderzoeken. De patiëntenverenigingen Duchenne Parent Project en Vereniging Spierziekten Nederland wil ik daarnaast graag bedanken voor hun bereidheid mij in contact te brengen met deze jongens/mannen en het actief meedenken over de invulling en uitvoering van mijn onderzoek.

Naast deze belangrijke groepen, wil ik ook graag enkele mensen persoonlijk bedanken. In de eerste plaats mijn promotor, Jan Verschuuren: jouw enthousiasme en optimisme werkten aanstekelijk, zeker op de momenten dat mijn van nature wat meer bedachtzame en sceptische aard de overhand leek te krijgen. Mijn co-promotor, Annemieke Aartsma: zonder jouw begrijpelijke uitleg over de wereld van het RNA en het exon skippen was ik waarschijnlijk nooit tot dit resultaat gekomen. Je kreeg het voor elkaar de meest complexe processen en systemen begrijpelijk te maken. To Eric Hoffman I would like to say thank you for providing me with the opportunity to explore the fascinating world of muscular dystrophy. I felt honored to work in your laboratory and think back to my stay in Washington with great pleasure. Fieke, Beatrijs en Zaïda, mijn onderzoeks-collega's, wat heb ik veel aan jullie gehad! In goede tijden kon ik mijn enthousiaste verhalen en resultaten met jullie delen en wanneer het even niet goed liep, boden jullie een luisterend oor en een bemoedigend woord. Gelukkig was er ook tijd voor niet onderzoek gerelateerde gesprekken. Nienke, ruim 9 jaar geleden begonnen we samen als arts-assistent neurologie in het Rijnland Ziekenhuis. Het was fijn iemand te hebben om alle nieuwe indrukken mee te delen. Hoewel we tijdens de opleiding grotendeels in verschillende ziekenhuizen werkten, voelde het altijd vertrouwd om je weer te spreken. En tot slot Kris, wat fijn dat je voor mij de cover hebt willen ontwerpen. Geduldig heb je keer op keer zaken uitgelegd en toegelicht, aanpassingen gemaakt als ik weer wat nieuws bedacht en adviezen gegeven over de opmaak van het proefschrift. Ik ben ontzettend blij met het eindresultaat!

Bij het tot stand komen van dit proefschrift was ook goede steun van het thuisfront onontbeerlijk. In de eerste plaats wil ik mijn moeder, Constance bedanken. Mam, je hebt me altijd meegegeven dat ik alles kan waar ik me voor inzet. Jouw voorbeeld: een intelligente, hardwerkende en warme vrouw, is inspirerend en je steun en oprechte interesse gedurende dit hele proces gaven me het vertrouwen dat ik het tot een goed einde zou kunnen brengen.

Tot slot een woord tot mijn lieve gezin: Robert, wat hebben we vaak gezellig samen aan de eettafel achter onze laptops gezeten! Maar wat heb jij ook veel geduld moeten hebben als ik je weer riep omdat ik ruzie had met een computerprogramma. Je gaf me de ruimte dit hele proces te ervaren en bood me de rust die ik hierbij nodig had.

Lieve Babette, Quirine en Elias, dit traject ben ik in 2007 gestart, ver voordat jullie er waren. Hoewel de weg naar een promotie een mooi en uniek proces is, ben ik blij dat het niet bij de geboorte van een proefschrift is gebleven en ik nu zulke gezellige, nieuwsgierige en bij vlagen ondeugende lieverds in mijn leven heb!

