



Universiteit
Leiden
The Netherlands

The use of new technology to improve genetic testing

Almomani, R.

Citation

Almomani, R. (2013, June 19). *The use of new technology to improve genetic testing*. Retrieved from <https://hdl.handle.net/1887/20981>

Version: Corrected Publisher's Version

License: [Licence agreement concerning inclusion of doctoral thesis in the Institutional Repository of the University of Leiden](#)

Downloaded from: <https://hdl.handle.net/1887/20981>

Note: To cite this publication please use the final published version (if applicable).

Cover Page



Universiteit Leiden



The handle <http://hdl.handle.net/1887/20981> holds various files of this Leiden University dissertation

Author: Almomani, Rowida

Title: The use of new technology to improve genetic testing

Issue Date: 2013-06-19

List of publications

Almomani R, van der Stoep N, Bakker E, den Dunnen JT, Breuning MH, Ginjaar IB. Rapid and cost effective detection of small mutations in the DMD gene by high resolution melting curve analysis. *Neuromuscul Disord*. 2009; 19:383-90.

Almomani R, van der Heijden J, Ariyurek Y, Lai Y, Bakker E, van Galen M, Breuning MH, den Dunnen JT. Experiences with array-based sequence capture; toward clinical applications. *Eur J Hum Genet*. 2011; 19: 50–55.

Sun Y*, **Almomani R***, Aten E, Celli J, van der Heijden J, Venselaar H, Robertson SP, Baroncini A, Franco B, Basel-Vanagaite L, Horii E, Drut R, Ariyurek Y, den Dunnen JT, Breuning MH. Terminal Osseous Dysplasia is Caused by a Single Recurrent Mutation in the *FLNA* Gene. *Am J Hum Genet*. 2010; 87:146-53. *The authors contributed equally to this work

Yu Sun*, **Rowida Almomani***, Guido Breedveld, Gijs W.E. Santen, Emmelien Aten, Dirk J. Lefeber, Jorrit I. Hoff, Esther Brusse, Frans W. Verheijen, Rob M. Verdijk, Marjolein Kriek, Ben Oostra, Martijn H. Breuning, Monique Losekoot, Johan T. den Dunnen, Bart P. van de Warrenburg, and Anneke J.A. Maat-Kievit. Autosomal recessive spinocerebellar ataxia 7 (SCAR7) is caused by variants in *TPP1*, the gene involved in classic late-infantile neuronal ceroid lipofuscinosis 2 disease (CLN2 disease). *Hum mutat*. 2013;34:706-13. *The authors contributed equally to the work

Rowida Almomani*, Yu Sun*, Emmelien Aten, Yvonne Hilhorst-Hofstee, Cacha M.P.C.D. Peeters-Scholte, Arie van Haeringen, Yvonne M.C. Hendriks, Johan T. den Dunnen, Martijn H. Breuning, Marjolein Kriek, and Gijs W.E. Santen. GPSM2 and Chudley–McCullough Syndrome: A Dutch Founder Variant Brought to North America. *Am J Med Genet Part A*. 2013. *The authors contributed equally to this work

Lemmers RJ, Tawil R, Petek LM, Balog J, Block GJ, Santen GW, Amell AM, van der Vliet PJ, **Almomani R**, Straasheijm KR, Krom YD, Klooster R, Sun Y, den Dunnen JT, Helmer Q, Donlin-Smith CM, Padberg GW, van Engelen BG, de Greef JC, Aartsma-Rus AM, Frants RR,

de Visser M, Desnuelle C, Sacconi S, Filippova GN, Bakker B, Bamshad MJ, Tapscott SJ, Miller DG, van der Maarel SM. Digenic inheritance of an *SMCHD1* mutation and an FSHD-permissive D4Z4 allele causes facioscapulohumeral muscular dystrophy type 2. *Nat Genet.* 2012 44:1370-4.

Santen GW, Aten E, Sun Y, **Almomani R**, Gilissen C, Nielsen M, Kant SG, Snoeck IN, Peeters EA, Hilhorst-Hofstee Y, Wessels MW, den Hollander NS, Ruivenkamp CA, van Ommen GJ, Breuning MH, den Dunnen JT, van Haeringen A, Kriek M. Mutations in SWI/SNF chromatin remodeling complex gene *ARID1B* cause Coffin-Siris syndrome. *Nat Genet.* 2012; 44:379-80.

Aten E*, Sun Y*, **Almomani R**, Santen GW, Messemaker T, Maas SM, Breuning MH, den Dunnen JT. Exome Sequencing Identifies A Branch Point Variant in Aarskog-Scott Syndrome. *Hum Mutat.* 2013;34:430-4. *The authors contributed equally to this work

B. Sikkema-Raddatz, L. F. Johansson, E. N. de Boer, **R. Almomani**, L.G. Boven, M.P. v.d. Berg, K.Y. van Spaendonck-Zwarts, JP van Tintelen, R. Sijmons, J.D.H. Jongbloed, R.J. Sinke. Targeted next generation sequencing ready for clinical diagnostics: Sanger sequencing can be replaced. *Hum Mutat.* 2013

Karin Y. van Spaendonck-Zwarts, Anna Posafalvi, Denise Hilfiker-Kleiner, Karen Sliwa, Mariel Alders, **Rowida Almomani**, Dirk J. Van Veldhuisen, Irene M. van Langen, Richard J. Sinke, Jolanda van der Velden, Maarten P. van den Berg, J. Peter van Tintelen, Jan D.H. Jongbloed. Targeted next generation sequencing in families with peripartum cardiomyopathy and dilated cardiomyopathy. submitted

Acknowledgements

I would like to thank all the people who have contributed to finish this thesis. It would have never have reached this point without their support, encouragement and help.

First, I would like to thank my promoters Prof. Martijn Breuning and Prof. Bert Bakker who gave me the opportunity to work as a PhD student in the Clinical and Human Genetic Department of the LUMC. Thank you for your kindness, support, patient guidance as well as your academic experience, which have been greatly valuable to me.

I would like to express my thanks to Ieke, for her personal support and great patience at all times, you are just a close family member who cares about me and who is always there.

Many thanks to Kamlesh for critical reading of my thesis and precise sense of the English language contributed to the final draft.

Johan, thanks for your kind assistance and willingness to discuss ideas about different projects.

Marjolein, Emmelien, Yu and Gijs, it was nice to work with all of you, so enjoyable and productive.

Many thanks to my colleagues at LDGA, working at this department has definitely became easier due to the nice and relaxing social environment, all of you were willing to help and answering my questions.

I would like to thank Astrid and Janina for shearing the same office with me. You are one of my best friends. You were nice company.

I want to thank the colleagues in LGTC. Yavuz, you are so smart, you are always willing to help, explain the experiments technical details in so easy way. I still need to learn from you, and still remember your words (no worries, who cares, details, tomorrow is another day). Thanks a lot for your help!

I would like to thank Rolf for introducing the melting curve analysis to me at the beginning of my PhD period. It was nice to work with you all the time at the LGTC.

I would like to thank also my colleagues at my work at the Department of Genetics, University of Groningen, University Medical Centre Groningen (UMCG), the Netherlands. It is indeed a nice place to work in, thanks Richard and Jan for your nice support.

I would like to express my thanks and love to my parents, brothers and my sister; they gave me the courage and support in my life.

In the end, I lack words to express my thanks, feelings, and love to my husband Ali and my two little angels Tala and Yara: you are the source of happiness, peace and love in my life, you were so patient during my PhD period, without your support I would have never finished my work. I was so busy during my PhD work, I know you missed me so much I was seeing that in your eyes, every day I came home and every weekend I was working in the lab. I love you my little family.

Curriculum Vitae

Rowida Almomani was born on 4th of February, 1979 in Al-Ruseifa, Jordan. She passed the general secondary education examination (Tawjihi) in 1997. She then studied Biology at Mutah University in Al-Karak, Jordan, and got her Bachelor degree with the highest average and thus being the top student at the Bachelor program for that year, 2001. She got the Honoree certificate for advanced academic achievements in the Bachelor period. In 2005, Rowida got her Master degree in applied biology from the Jordan University of Science and Technology (JUST) in Irbid, Jordan. Two years later she joined the group of Prof. Martijn H Breuning, at the Clinical and human genetics department of Leiden University Medical Center (LUMC) in Leiden, the Netherlands, to do her PhD. During her PhD work, she has been introduced to different molecular diagnostic technologies including the rapidly developing field of next generation sequencing (NGS). Her research focuses on the application of NGS and the use of new technologies, especially High Resolution Melting Curve Analysis (HR-MCA), targeted and exome sequencing, to be able to identify the causal pathogenic variants in different genetic diseases and to improve genetic testing. In December 2011 until now, she works as a post doc at the department of Genetics of the University Medical Center Groningen (UMCG), the Netherlands. Currently her research subject focuses on finding pathogenic mutations in genes related to Cardiomyopathies and Heart diseases by exome sequencing. The research projects she worked with have provided her with a broad view that makes her able to work independently as well as in a team-work and makes her highly motivated to work in different areas of genetic research.