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## The use of new technology to improve genetic testing

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## 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.

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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**

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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,

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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

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## **Curriculum Vitae**

Rowida Almomani was born on 4<sup>th</sup> 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.