Dr. Rowida Almomani
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Education

- **PhD in human genetics From Leiden University, the Netherlands (19-06-2013)**
  - PhD Thesis: **The use of new technology to improve genetic testing**
- **Master degree in applied biology, genetics (accumulative average of 85.5%, 2002-2005), Jordan University of science and Technology, Jordan**
  - Master Thesis: **Screening for common mtDNA mutations especially for those causing MERRF, MELAS disorders among Jordanian population**
- **Bachelor degree in Biology (accumulative average of 85.7%, excellent, 1997 – 2001), Mutah University, Jordan**
- **High school degree in 1997, Jordan (Average 82.5%)**

Work experience

- March 2017-present, Assistant Professor in the Department of Medical Laboratory Sciences, Jordan University of Science and Technology
- January 2015- April 2017, Team member of Neuromuscular Diseases Center Maastricht (Spierziekten Centrum Maastricht), The Netherlands
- January 2015- April 2017, work Package (WP2) leader in an international collaborative project, the PROPANE* study
- May 2014 – April 2017, postdoctoral researcher at the department of human genetics and cell biology, Maastricht University, The Netherlands
- January 2012- April 2014, postdoctoral researcher at the department of human genetics, University Medical Center Groningen (UMCG), University of Groningen
- Teaching assistant at the department of biology, Jordan University of Science and Technology, 2002-2005
- Teacher at Yarmouk University Model School, February- June 2002

* PROPANE study: an international collaborative project with colleagues from Department of Clinical Genetics and Department of Neurology, University Medical Center, Maastricht, the Netherlands; Neuromuscular Diseases Unit and Bioinformatics Unit, IRCCS Foundation,

"Carlo Besta", Milan, Italy; Department of Neurology, Spaarne Hospital, Hoofddorp, the Netherlands; Laboratory of Genetics of Neurological Complex Disorders, San Raffaele Scientific Institute, Milan, Italy; Department of Neurology and Center for Neuroscience and Regeneration Research, Yale University School of Medicine, New Haven.

Publications

1. Martinelli-Boneschi F, Colombi M, Castori M, Devigili G, Eleopra R, Malik RA, Ritelli M, Zoppi N, Dordoni C, Sorosina M, Grammatico P, Fadavi H, Gerrits MM, **Almomani R**, Faber CG, Merkies IS, Toniolo D; INGI Network, Cocca M, Doglioni C, Waxman SG, Dib-Hajj SD, Taiana MM, Sassone J, Lombardi R, Cazzato D, Zauli A, Santoro S, Marchi M, Lauria G. COL6A5 variants in familial neuropathic chronic itch. Brain. 2017 Mar 1;140(3):555-567. **Impact Factor: 10.2**

2. **Almomani R***, Verhagen JM*, Herkert JC, Brosens E, van Spaendonck-Zwarts KY, Asimaki A, van der Zwaag PA, Frohn-Mulder IM, Bertoli-Avella AM, Boven LG, van Slegtenhorst MA, van der Smagt JJ, van IJcken WF, Timmer B, van Stuijvenberg M, Verdijk RM, Saffitz JE, du Plessis FA, Michels M, Hofstra RM, Sinke RJ, van Tintelen JP, Wessels MW, Jongbloed JD*, van de Laar IM*. **Biallelic truncating mutations in *ALPK3* cause pediatric cardiomyopathy. *J Am Coll Cardiol*. 2016;67:515-525. The authors contributed equally to this work. Impact Factor: 16.5**

3. van Spaendonck-Zwarts KY, Posafalvi A, van den Berg MP, Hilfiker-Kleiner D, Bollen IA, Sliwa K, Alders M, **Almomani R**, van Langen IM, van der Meer P, Sinke RJ, van der Velden J, Van Veldhuisen DJ, van Tintelen JP, Jongbloed JD. Titin gene mutations are common in families with both peripartum cardiomyopathy and dilated cardiomyopathy. **European Heart Journal.** 2014;35:2165-73. **Impact Factor: 15.20**

4. 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 can replace Sanger sequencing in clinical diagnostics. **Human Mutation**. 2013;34:1035-42**. Impact Factor:** 5.34

5. Sun Y*, **Almomani R***, Breedveld GJ, Santen GW, Aten E, Lefeber DJ, Hoff JI, Brusse E, Verheijen FW, Verdijk RM, Kriek M, Oostra B, Breuning MH, Losekoot M, den Dunnen JT, van de Warrenburg BP, Maat-Kievit AJ. 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). **Human mutation**, 2013; 34:706-13. ***The authors contributed equally to the work. Impact Factor:** 5.34

6. **Almomani R*,** Sun Y* Aten E, Hilhorst-Hofstee Y, Peeters-Scholte CM, van Haeringen A, Hendriks YM, den Dunnen JT, Breuning MH, Kriek M, Santen GW. GPSM2 and Chudley–McCullough Syndrome: A Dutch Founder Variant Brought to North America. American Journal of Medical Genetics Part A. **2013;161A:973-6. *The authors contributed equally to this work. Impact Factor: 2.16**

7. 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. **Human Mutation**. 2013;34:430-4.***The authors contributed equally to this work. Impact Factor:** 5.34

8. 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. **Nature Genetics**. 2012 44:1370-4. **Impact Factor: 29.35**

9. 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. **Nature Genetics**. 2012; 44:379-80. **Impact Factor: 29.35**

10. **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. **European Journal of Human Genetics.** 2011; 19: 50–55. **Impact Factor: 4.35**
11. **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. **The American Journal of Human Genetics**, 2010, 87(1), 146-153. ***The authors contributed equally to this work. Impact Factor: 10.93**

12. **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. **Impact Factor: 2.64**

Manuscripts in preparation

1. **Almomani R***, Pósafalvi A*, Post JG, van der Zwaag PA, Corsten-Janssen N, Niezen KE, de Koning TJ, Rodenburg RJ, Sinke RJ, van Tintelen JP, Jongbloed JD. Exome sequencing identifies a mutation in the nuclear encoded mitochondrial protein ***X*** causing cardiomyopathy. ***These authors contributed equally; to be submitted.**

2. **van den Berg MP***, **Almomani R*,** Biaggioni I, van der Harst P, Jongbloed JDH, Herman HW Sillje, Hemmelder MH, Navis G, Luijckx GJ, Wever RA, van Tintelen JP, Kema IP. A hereditary orthostatic hypotension syndrome due mutations in ***X*** gene. **Submitted.**

3. **Almomani R***, Pósafalvi A, van der Zwaag PA, Lazzarini E, Boven LG, Abbott K, van Tintelen JP, Sinke RJ, Jongbloed JD. Combining Exome sequencing and Haplotype Sharing Test for cardiomypathy disease gene discovery. **Manuscript in preparation**

4. **Rowida Almomani**, Patrick Lindsey, Catharina G. Faber, Paola Tononi, Giuseppe Lauria, Ingemar S.J. Merkies, Janneke G.J. Hoeijmakers, Bianca de Greef, Maurice Sopacua, Hubert Smeets, Filippo Martinelli Boneschi, Sulayman Dib-Hajj, Stephen G Waxman, Monique M. Gerrits; PROPANE Study Group. **Molecular Inversion Probe-targeted Next Generation Sequencing to Identify Genetic Variations. Manuscript in preparation**

Expertise in diagnostic settings

- I optimized and validated HR-MCA method to scan all coding exons of the DMD gene in diagnostic settings. The method is implemented in routine diagnostic use on patients with Duchenne or Becker muscular dystrophy and on female carriers (Leiden University Medical Center/ LUMC/ The Netherlands)
- During my postdoc period in UMCG, I participated in validating targeted next generation sequencing (in-solution custom made kit) to capture and sequence 55 genes known to be associated with cardiomyopathy. Currently, this method has been implemented as a diagnostic test for cardiomyopathy patients (University Medical Center Groningen (UMCG), the Netherlands)
- I optimized and validated molecular inversion probes technique followed by next generation sequencing (MIPs-NGS) to be implemented in diagnostic to screen patients with neuropathic pain (University Medical Center, Maastricht University, the Netherlands)

Technical Expertise

- **Extensive experience of Next generation sequencing (NGS) technologies and assays:**
  - Sample preparation for Illumina (Hiseq, Miseq, and Nextseq), **DNA fragmentation** using nebulization, sonication (biorupter) and by Covaris.

*** Capture by Circularization**:

- **Molecular Inversion Probe (MIP):** Develop, validate and implement MIPs approach to capture specific genomic regions

* **Capture by Hybridization:**

- **Solid capture**: design and use microarrays (2.1 million whole exome and 385kb custom arrays) to capture genomic regions

- **In-solution capture**: Whole exome/ X-chromosome/ custom made kits (design probes) to capture genomic regions of interest (Agilent and Nimblegen)

- **Next generation sequencing (NGS) data analysis:**

  - Familiar with using various sequence analysis tools, including BWA, GATK, Picard, samtools and using R language.

  - Experienced in NGS data analysis by ClC bio-software and nextgen software

  - Extensive experience in NGS data visualization by using genome browsers such as UCSC, Alamut software and Integrative Genomics Viewer (IGV)

  - Extensive experience in analyzing NGS data using established workflows, evaluate data quality (QC measures) and apply analysis approaches to aid variant prioritization

  - Highly experienced in disease-focused genomics projects

  - Experienced in Genetic variations classification

  - Experience with commonly used public genomics datasets (1000 genomes, ExAC, OMIM, ClinVar, HGMD, GoNl (Genome of the Netherlands), NHLBI GO Exome Sequencing Project (ESP) etc.)

- **Extensive experience in PCRs** (nested PCR, Long range PCR, Real time PCR, RT-PCR, Primes and probes design)
- **Extensive experience in Microarrays** (CGH arrays and custom arrays) for copy number variation detection (CNVs/ deletions and duplications)
- **Extensive experience in Gel electrophoresis,** polyacrylamid gel , gel documentation system, DNA fragment size selection, gel extraction and purification
- **Extensive experience in High resolution melting analysis (HRMCA)**
- **Extensive experience in DNA and RNA isolation and nucleic acid concentration measurements** (Nanodrop, bio-analyzer from Agilent and Qubit assay)and **cDNA synthesis**

- **Extensive experience in Sanger sequencing and data analysis**
- Familiar with western blot
- Familiar with cell culture (fibroblast, fibroma tissue, bacteria)
- Familiar with exon trapping technique
- Experienced in Mononuclear cells isolation
- Experienced in Multiplex ligation-dependent probe amplification (MLPA)
- **Experienced in Writing scientific articles, documents and research reports (Preparation of summary, abstracts, conferences presentation, as well as publications based on the conducted research)**
- **Writing grants proposal**
- **Guide and supervise researchers, technicians & undergraduate students**
- **Designing and participate in designing courses, research and experiments**

Meetings and Courses

- **PROPANE STUDY Annual Meeting, October 2015,** Belgium**.**

  Oral presentation: Targeted and unbiased genetic analyses.
- **Peripheral Nerve Society meeting, June 2015, Canada.**
- **Poster presentation:** Molecular Inversion Probe-targeted Next Generation Sequencing to Identify Genetic Causes of Painful Neuropathy.
- **ICIN- Netherlands Heart Institute meeting, 2013, the Netherlands.**

  Oral presentation: Identification of disease genes causing cardiomyopathies by exome sequencing
- **Invited speaker at ESHG conference 2013, Agilent Technologies, France.**

  Oral presentation: Targeted resequencing results in a significant increase in identifying genetic causes of cardiomyopathies.
- **American heart association conference (AHA) 2012, USA.**

  Poster presentation: Targeted and Exome Sequencing and Haplotype Sharing Analyses Result in the Identification of Novel Genetic Causes of Cardiomyopathies
- **ESHG conference, 2012. Poster presentation:** Whole-exome sequencing identifies a novel nonsense mutation in the TTN gene in a large Dutch family with DCM

- **NVGH meeting 2012, The Netherlands. Oral presentation:** Targeted Next Generation Resequencing in Cardiomyopathy Patients Can Substitute Sanger Sequencing and Results in a Significant Increase in Diagnostic Yield.
- **NMD chip meeting, 2011, London. Oral presentation**.
- **ESHG conference 2011, The Netherlands. Poster presentation.**
- **NVGH meeting 2010, the Netherlands. Poster presentation:** Targeted DNA-CGH arrays for high throughput diagnosis of muscular dystrophies.
- **ESHG conference 2010, Sweden. Poster presentation.**
- **NVGH meeting 2009, the Netherlands. Poster presentation**
- **ASHG 2009, USA, poster presentation**: Reliable recovery of mutations and CNVs (incl. all known) using array-based sequence capture of 112 MR-genes.
- **NMD chip meeting, 2009, France. Oral presentation.**

Courses and Teaching

- I organized and gave a training course to learn the 'Molecular Inversion Probe (MIP) based targeted enrichment technique', December 2015, at University Medical Center, Maastricht, and The Netherlands.
- **Invited lecturer at Avans Hgeschool, Breda, the Netherlands.** Next Generation Sequencing; Technologies, applications and data analysis (5-8 April 2011/ Avans Hgeschool, Breda)
- **Special KNAW Conference (and Master classes):** From DNA variations to phenotype, 9 - 11 March 2011, Rotterdam
- I organized and gave a training course to learn the micro-array-based targeted enrichment protocol. **Leiden university medical center, Leiden, the Netherlands, 2010.**
- **Invited lecturer at Avans Hgeschool, Breda, the Netherlands**. Next generation sequencing; Technologies, applications and data analysis (February 9-12 2010, guest lecturer / Avans Hgeschool, Breda)
- **Invited lecturer at Leiden university medical center, Leiden, the Netherlands, 2010**. Next generation sequence data analysis, March 17 - 19, 2010 (attending and giving a lecture/ guest lecturer)
- Epigenetic Regulation (2010 Leiden)

- **Invited lecturer at Leiden university medical center, Leiden, the Netherlands, 2009.** Iintroduction to Next Generation Sequencing; technologies and data analysis at (attending the whole course and giving a lecture)
- **Invited lecturer at Technology Facilities at Leiden university medical center, Leiden (LUMC)** (attending and giving a lecture)
- Intensive R Course for Micro-array Data Analysis
- From development to disease, 2007, Erasmus MC, Rotterdam
- Introduction to Linux course
- Advanced statistic course 2007

Teaching courses at Jordan University of Science and Technology:

- ADVANCED MOLECULAR BIOLOGY
- ADVANCED HUMAN CYTOGENETIC
- SPECIAL TOPICS IN HUMAN GENETICS
- ADVANCED HUMAN GENETICS
- MOLECULAR BIOLOGY
- DIAGNOSTIC MOLECULAR BIOLOGY AND CYTOGENETIC

Honorees and Awards

- Trainee travel fellowship (**Peripheral Nerve Society meeting, 2015, Canada)**
- Scholarship for the PhD study from Leiden University, the Netherlands.
- Scholarship for the master study from Jordan University of science and technology.
- The highest average to be the top student in the Bachelor program (1997 – 2001).
- I got Honoree certificate for advanced academic achievements in the BSc

Research interests

- The application of genomic technologies to understand the basis of different genetic disorders.
- Exploring the latest genomic technologies for clinical/diagnostic use in many genetic disorders, including cardiomyopathies, painful neuropathy, mental retardation, and muscle diseases

References

- Prof. Dr. J.P. van Tintelen

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- Dr. Jan Jongbloed

Email address: j.d.h.jongbloed@umcg.nl

- Dr. Monique Gerrits

Email address: monique.gerrits@mumc.nl

Dr. Ieke Ginjaar

Email address: H.B.Ginjaar@lumc.nl

- Prof. Dr. M.H. Breuning

Email address: m.h.breuning@lumc.nl