Curriculum Vitae

Dr. Mohammad Abdullah Al-shboul, Ph.D.

Assistant Professor
Human Genetics and Molecular Biology
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Personal Information:

Date of birth : 13th June, 1979

Gender : Male

Nationality : Jordanian

Marital status : Married

Goals:

1) Understand the molecular mechanisms underlying rare genetic disorders.

2) Develop new and reliable methods for molecular diagnosis of patients with genetic disorders. This will help them receive appropriate therapy and for their families to plan for future healthy children

3) Along with research, I am also interested in teaching scientists and undergraduate or graduate students, sharing my knowledge with them and giving them the best of my abilities. I plan on keep this interplay between research diagnostic and teaching in the future.

Education:

2010-2014 Ph.D., National University of Singapore, Yong Loo Lin School of Medicine, Singapore.

1997-2002 Bachelor Degree in Medical Laboratory Sciences, School of Applied Medical Science, Jordan University of Science and Technology, Jordan.

Professional Experience:

2018-Present Assistant Professor, Department of Medical Laboratory Sciences, Jordan University of Science and Technology, Irbid 22110, Jordan

2017-2018 Medical Laboratory Sciences, Faculty of Science, Al-Balqa' Applied University, Al-Salt - Jordan

2014-2017 Postdoctoral Research Fellow at the Institute of Medical Biology (IMB), Human Embryology and Genetics Laboratory (*June 2014*), Singapore.

2010-2014 Ph.D. research work at IMB, Human Embryology and Genetics Laboratory, Singapore. *The degree was conferred on November, 2014.*

2005-2009 Part-time Senior Molecular Geneticist at the Specialty Centre for Fertility and Genetics, Al-khaldi Medical Centre, Jordan.

2003-2009 Laboratory Supervisor of the genetic laboratory, University of Jordan and National Centre for Diabetes, Endocrinology and Genetics, Jordan.

Responsibilities

- Management of the Genetic Lab resources: ordering and purchasing of instruments, reagents of cytogenetic and molecular genetic laboratories.
- Trained of graduate/undergraduate students and lab technicians from other institutions in Jordan as well as from other countries on molecular genetics techniques.
- Applying of the following basic procedures: DNA/RNA extraction from various samples: blood, saliva, chorionic villus samples (CVS), amniocytes, paraffin-embedded tissue and others, DNA/RNA quantification, optimization of polymerase chain reaction (PCR) and results interpretation.
- Applying various molecular biology techniques including: Amplification of Refractory Mutation System (ARMS-PCR), PCR-Restriction Enzyme Analysis (PCR-RE), Restriction Fragment Length Polymorphisms (RFLPs), Variable Number of Tandem Repeats (VNTRs), DNA Sequencing, Fragment Analysis and Quantitative Fluorescent PCR (QF-PCR).
- Sanger sequencing and fragment analysis for genetic disorders
- Direct mutation detection for genetic disorders such as: Familial Mediterranean Fever (FMF) β & α Thalassemia, Coagulation Factors, MTHFR, Cystic Fibrosis (CF), Hereditary Hemochromatosis, α-1 antitrypsin, Achondroplasia, Mitochondrial, Sanjad-Sakati and other disorders.
- Molecular testing for: Angiotensin Converting Enzyme 1 (ACE), Sex Determining Region (SRY), Sex Chromosome Determination (AMXY), Azospermia, Muscle diseases including Duchenne Muscular Dystrophy (DMD), Spinal Muscular Atrophy (SMA) and others, Apolipoproteine E genotyping and Human Leukocyte Antigen (HLA) typing and other single gene disorders: Phenylketonuria (PKU), Hemophilia and Tri-nucleotides repeat expansion diseases such as Myotonic Dystrophy type 1 (DM1), Spinocerebellar Ataxia (SCA), Friedreich's Ataxia (FRDA) and Huntington.
- Prenatal diagnosis testing.
- Next generation sequencing: Post-computational analysis including: Inheritance pattern analysis, in silico analysis using bioinformatics tools and validation of identified variants, etc.

2002-2004 Laboratory Technician in different medical laboratories (Hematology, Clinical Chemistry, Endocrinology, Serology, & Microbiology), Amman, Jordan.

Ph.D. Research

In general, we aim to identify genes responsible for rare monogenic disorders and investigate their function in the context of normal and disease state. Studying these unique diseases provide us with invaluable insights into normal physiology and give us a better understanding of biological pathways that could be similarly affected in more common ailments. In essence, we believe that rare begets common. My work during Ph.D. focused on studying the genetic aetiology of rare ciliopathies known as Oral-facial-digital syndromes (OFD) particularly OFDII or Mohr syndrome. Harnessing the power of SNP-mapping and next-generation sequencing analyses (WES and WGS), I uncovered the genetic lesion responsible for this disease. Using *in vitro* assays with patient's cells and *in vivo* animal models, I have documented the role of the causative gene in cilia formation and function and that its biallelic disruption in humans causes Mohr Syndrome.

I also participated in identification of genetic lesions of many ciliopathies and other congenital disorders.

Research Experience and Technical Expertise:

- Next generation sequencing: Post-computational analysis including: Inheritance pattern analysis, *in silico* analysis using bioinformatics tools and validation of identified variants, etc.
- Cell culture and maintenance for a variety of cell types.
- Transient and stable transfection of different cell lines.
- Generation of human induced pluripotent stem cells (iPSCs) and differentiating them into neural cells and other cell types.
- DNA / RNA techniques: Plasmid DNA isolation, molecular cloning, RNA isolation, primer design, PCR, quantitative real time-PCR, RACE-PCR, site-directed mutagenesis and gel electrophoresis, etc.
- Protein techniques: Protein extraction, western blot and immunoprecipitation, etc.
- Functional analysis:
  - Gene silencing: siRNA approach, shRNA and CRISPR.
  - Frog and zebrafish husbandry and manipulation.
  - *In situ* hybridization and Immunocytochemistry for cells and embryos.

Award Scholarships:

2010-2014 Singapore International Graduate Award (SINGA), A*STAR.

2009-2009 Singapore International Pre-Graduate Award (SIPGA), A*STAR, for 6 months.

Conferences Participation and Other Activities:

- The 8th *Pan Arab Human Genetics Conference (PAHGC)*, Dubai, January, 2020.
- Cilia 2016, *from fundamental biology to human disease*, Amsterdam, The Netherlands October, 2016,
- The 6th *Pan Arab Human Genetics Conference (PAHGC)*, Dubai, January, 2016.
- The 5th *Pan Arab Human Genetics Conference* (*PAHGC*), Dubai, November, 2013.
- The '*Hedgehog Signaling in Development Evolution and Disease*, *Singapore*, *March*, 2012.
- DNA sequencing on Beckman-Coulter sequencer (CEQ8000) at Cyprus Institute for Neurogenetics, July 2005.
- The final year of the Bachelor Degree was a training year (24 credit hours). I had an opportunity to explore and work in various labs at prince's Basma hospital to perform all routine and special tests such as Hematology, chemistry, serology, microbiology, histopathology and Blood Banking, From July (2001-2002).

Publications:

1. Nabavizadeh N, **Shboul M**, Hojati Z. Bioenergetic analysis of aged-phenotype skin in a rare syndromic cutis laxa. J Cosmet Dermatol. 2021 Feb 1. doi: 10.1111/jocd.13951. Epub ahead of print. PMID: 33522694.
2. Al Smadi MA, Hammadeh ME, Solomayer E, Batiha O, Altalib MM, Jahmani MY, **Shboul MA**, Nusair B, Amor H. Impact of Mitochondrial Genetic Variants in ND1, ND2, ND5, and ND6 Genes on Sperm Motility and Intracytoplasmic Sperm Injection (ICSI) Outcomes. Reprod Sci. 2021 Jan 21. doi: 10.1007/s43032-020-00449-3. Epub ahead of print. PMID: 33475980.
3. Bouchoucha S, Chikhaoui A, Najjar D, Dallali H, Khammessi M, Abdelhak S, Nessibe N, **Shboul M**, Kircher SG, Al Kaissi A, Yacoub-Youssef H. Clinical and Genetic Heterogeneity in Six Tunisian Families With Horizontal Gaze Palsy With Progressive Scoliosis: A Retrospective Study of 13 Cases. Front Pediatr. 2020 Apr 16;8:172. doi: 10.3389/fped.2020.00172. PMID: 32373565; PMCID: PMC7179758.
4. Kaissi AA, Kenis V, **Shboul M**, Grill F, Ganger R, Kircher SG. Tomographic Study of the Malformation Complex in Correlation With the Genotype in Patients With Robinow Syndrome: Review Article. J Investig Med High Impact Case Rep. 2020 Jan-Dec;8:2324709620911771. doi: 10.1177/2324709620911771. PMID: 32172608; PMCID: PMC7074505.
5. Al Kaissi A, Ryabykh S, Ochirova P, Bouchoucha S, Kenis V, **Shboul M**, Ganger R, Grill F, Kircher SG. Arthrogryposis is a descriptive term, not a specific disease entity: escobar syndrome is an Example. Minerva Pediatr. 2020 Jun 12. doi: 10.23736/S0026-4946.20.05796-5. Epub ahead of print. PMID: 32536119.
6. Trott J, Alpagu Y, Tan EK, **Shboul M**, Dawood Y, Elsy M, Wollmann H, Tano V, Bonnard C, Eng S, Narayanan G, Junnarkar S, Wearne S, Strutt J, Kumar A, Tomaz LB, Goy PA, Mzoughi S, Jennings R, Hagoort J, Eskin A, Lee H, Nelson SF, Al-Kazaleh F, El-Khateeb M, Fathallah R, Shah H, Goeke J, Langley SR, Guccione E, Hanley N, De Bakker BS, Reversade B, Dunn NR. Mitchell-Riley syndrome iPSCs exhibit reduced pancreatic endoderm differentiation due to a mutation in RFX6. Development. 2020 Nov 5;147(21):dev194878. doi: 10.1242/dev.194878. PMID: 33033118.
7. **Shboul M**, Masri A, Khasawneh A, Jadallah R, ALmustafa A, Escande-Beillard N Hamamy H, Bakri F, Reversade B. Congenital insensitivity to pain with anhidrosis syndrome: A series from Jordan. Clin Neurol Neurosurg. 2020 Feb;189:105636. doi: 10.1016/j.clineuro.2019.105636. Epub 2019 Dec 9. PubMed PMID: 31841741.
8. Al Kaissi A, Kenis V, Jemaa LB, Sassi H, **Shboul M**, Grill F, Ganger R, Kircher SG. Skeletal phenotype/genotype in progressive pseudorheumatoid chondrodysplasia. Clin Rheumatol. 2020 Feb;39(2):553-560. doi: 10.1007/s10067-019-04783-z. Epub2019 Oct 18. PubMed PMID: 31628567.
9. Albaramki J, Dmour H, **Shboul M**, Bonnard C, Venkatesh B, Odeh R. Recessive mutation in GALNT3 causes hyperphosphatemic familial tumoral calcinosis associated with chronic recurrent multifocal osteomyelitis. Turk J Pediatr. 2019;61(1):130-133. doi: 10.24953/turkjped.2019.01.022. PubMed PMID: 31559735.
10. Al Kaissi A, **Shboul M**, Kenis V, Grill F, Ganger R, Kircher SG. Leri-Weill Dyschondrosteosis Syndrome: Analysis via 3DCT Scan. Medicines (Basel). 2019 May 29;6(2). pii: E60. doi:

10.3390/medicines6020060. PubMed PMID: 31146331; PubMed Central PMCID: PMC6631815.

11. Kaissi AA, Bouchoucha S, **Shboul M**, Kenis V, Grill F, Ganger R, Kircher SG. Massive Axial and Appendicular Skeletal Deformities in Connection with Gorham-Stout Syndrome. Medicines (Basel). 2019 May 7;6(2). pii: E54. doi: 10.3390/medicines6020054. PubMed PMID: 31067823; PubMed Central PMCID: PMC6631250.
12. Tian J, Shao J, Liu C, Hou HY, Chou CW, **Shboul M**, Li GQ, El-Khateeb M, Samarah OQ, Kou Y, Chen YH, Chen MJ, Lyu Z, Chen WL, Chen YF, Sun YH, Liu YW. Deficiency of lrp4 in zebrafish and human LRP4 mutation induce aberrant activation of Jagged-Notch signaling in fin and limb development. Cell Mol Life Sci. **2018** Oct 16. doi: 10.1007/s00018-018-2928-3. [Epub ahead of print] PubMed PMID: 30327840.
13. **Shboul M**, Roschger P, Ganger R, Paschalis L, Rokidi S, Zandieh S, Behunova J, Muschitz C, Fahrleitner-Pammer A, Ng AYJ, Tohari S, Venkatesh B, Bonnard C, Reversade B, Klaushofer K, Al Kaissi A. Bone matrix hypermineralization associated with low bone turnover in a case of Nasu-Hakola disease. Bone. **2018** Oct 10. pii: S8756-3282(18)30372-7. doi: 10.1016/j.bone.2018.10.008. [Epub ahead of print] PubMed PMID: 30316000.
14. Kou Y, **Shboul M**, Wang Z, Shersheer Q, Lyu Z, Liu P, Zhao X, Tian J. Novel frame shift mutation in ERCC6 leads to a severe form of Cockayne syndrome with postnatal growth failure and early death: A case report and brief literature review. Medicine (Baltimore). **2018** Aug;97(33):e11636. doi: 10.1097/MD.0000000000011636. Review. PubMed PMID: 30113454; PubMed Central PMCID: PMC6112894.
15. Chia PH, Zhong FL, Niwa S, Bonnard C, Utami KH, Zeng R, Lee H, Eskin A, Nelson SF, Xie WH, Al-Tawalbeh S, El-Khateeb M, **Shboul M**, Pouladi MA, Al-Raqad M, Reversade B. A homozygous loss-of-function CAMK2A mutation causes growth delay, frequent seizures and severe intellectual disability. Elife. **2018** May 22;7. pii: e32451. doi: 10.7554/eLife.32451. PubMed PMID: 29784083; PubMed Central PMCID: PMC5963920.
16. Bonnard C, **Shboul M**, Tonekaboni SH, Ng AYJ, Tohari S, Ghosh K, Lai A, Lim JY, Tan EC, Devisme L, Stichelbout M, Alkindi A, Banu N, Yüksel Z, Ghoumid J, Elkhartoufi N, Boutaud L, Micalizzi A, Brett MS, Venkatesh B, Valente EM, Attié-Bitach T, Reversade B, Kariminejad A. Novel mutations in the ciliopathy-associated gene CPLANE1 (C5orf42) cause OFD syndrome type VI rather than Joubert syndrome. Eur J Med Genet. 2018 Mar 30. pii: S1769-7212(17)30410-X. doi: 10.1016/j.ejmg.2018.03.012.
17. Yamauchi T, Masuda T, Canver MC, Seiler M, Semba Y, **Shboul M**, Al-Raqad M, Maeda M, Schoonenberg VAC, Cole MA, Macias-Trevino C, Ishikawa Y, Yao Q, Nakano M, Arai F, Orkin SH, Reversade B, Buonamici S, Pinello L, Akashi K, Bauer DE, Maeda T. Genome-wide CRISPR-Cas9 Screen Identifies Leukemia-Specific Dependence on a Pre-mRNA Metabolic Pathway Regulated by DCPS. Cancer Cell. 2018 Mar 12;33(3):386-400.e5. doi: 10.1016/j.ccell.2018.01.012. Epub 2018 Feb 22. PubMed PMID: 29478914; PubMed Central PMCID: PMC5849534.
18. Trott J, Tan EK, Ong S, Titmarsh DM, Denil SLIJ, Giam M, Wong CK, Wang J, **Shboul M**, Eio M, Cooper-White J, Cool SM, Rancati G, Stanton LW, Reversade B, Dunn NR. Long-Term Culture of Self-renewing Pancreatic Progenitors Derived from Human Pluripotent Stem Cells.

Stem Cell Reports. 2017 Jun 6;8(6):1675-1688. doi: 10.1016/j.stemcr.2017.05.019. PubMed PMID: 28591650; PubMed Central PMCID: PMC5470345.

19. Shifeng Xue, Jérôme Maluenda, Florent Marguet, **Mohammad Shboul**, Loïc Quevarec, Alvin Yu Jin Ng, Sumanty Tohari, Thong Teck Tan, Mung Kei Kong, Fawaz Alkazaleh, Marie Gonzales, Luc Rigonnot, Sandra Whalen, Marta Gut, Ivo Gut9, Martine Bucourt, Byrappa Venkatesh, Annie Laquerrière, Bruno Reversade and Judith Melki . Loss-of-function mutations in LGI4, encoding a secreted ligand involved in Schwann cell myelination, are responsible for arthrogryposis multiplex congenital. Am J Hum Genet. 2017 Apr 6;100(4):659-665.
20. Corey J Cain, Nathalie Gaborit, Wint Lwin, Emilie Barruet, Samantha Ho, Carine Bonnard, Hanan Hamamy, **Mohammad Shboul**, Bruno Reversade, Hülya Kayserili, Benoit G Bruneau, Edward C Hsiao **(2016)**. Loss of Iroquois Homeobox Transcription Factors 3 and 5 in Osteoblasts Disrupts Cranial Mineralization. Bone Reports, 2016, 5, 86–95.
21. Zhou F, Narasimhan V, **Shboul M**, Chong YL, Reversade B, Roy S **(2015)**: Gmnc Is a Master Regulator of the Multiciliated Cell Differentiation Program. Curr Biol 25, 24, 3267–3273.
22. **Mohammad Shboul**; Calista ng; Valerio Taverniti; Carine Bonnard; Hane Lee; Ascia Eskin; Stanley F Nelson; Mohammad Al-raqad; Samah Altawalbeh; Bertrand Seraphin; Bruno Reversade **(2014)**. Loss of the Scavenger mRNA Decapping Enzyme DCPS Causes Syndromic Mental Retardation with Neuromuscular Defects. Hum Mol Genet, 2015, 24(11): 3163–3171.
23. Wen F. Hu; Oz Pomp; Tawfeg Ben-Omran; Andrew Kodani; Katrin Henke; Ganeshwaran H. Mochida; Timothy W. Yu; Mollie B. Woodworth; Carine Bonnard; race Selva Raj; Thong Teck Tan; Hanan Hamamy; Amira Masri; **Mohammad Shboul**; Muna Al Saffar; Jennifer N. Partlow; Mohammed Al-Dosari; Anas Alazami; Mohammed Alowain; Fowzan S. Alkuraya; Jeremy F. Reiter; Matthew P. Harris; Bruno Reversade; Christopher A. Walsh **(2014)**. Katanin p80 regulates human cortical development by limiting centriole and cilia number. Neuron 84, 1–18.
24. Zimoń M1, Battaloğlu E, Parman Y, Erdem S, Baets J, De Vriendt E, Atkinson D, Almeida-Souza L, Deconinck T, Ozes B, Goossens D, Cirak S,Van Damme P, **Shboul M**, Voit T, Van Maldergem L, Dan B, El-Khateeb MS, Guergueltcheva V, Lopez-Laso E, Goemans N, Masri A, Züchner S, Timmerman V, Topaloğlu H, De Jonghe P, Jordanova A. **(2014)**. Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach. Neurogenetics. PubMed PMID: 25231362.
25. Jodoin, J.N., **Shboul, M**., Albrecht, T.R., Lee, E., Wagner, E.J., Reversade, B., and Lee, L.A. (**2013a**). The snRNA-processing complex, Integrator, is required for ciliogenesis and dynein recruitment to the nuclear envelope via distinct mechanisms. Biology open 2, 1390-1396.
26. Lopez E, Thauvin-Robinet C, Reversade B, Khartoufi NE, Devisme L, Holder M, Ansart-Franquet H, Avila M, Lacombe D, Kleinfinger P, Kaori I, Takanashi JI, Le Merrer M, Martinovic J, Noël C, **Shboul M**, Ho L, Güven Y, Razavi F, Burglen L, Gigot N, Darmency-Stamboul V, Thevenon J, Aral B, Kayserili H, Huet F, Lyonnet S, Le Caignec C, Franco B, Rivière JB, Faivre L, Attié-Bitach T. **(2013)**. C5orf42 is the major gene responsible for OFD syndrome type VI. Human genetics 133, 367-377.
27. Jodoin, J.N., Sitaram, P., Albrecht, T.R., May, S.B., **Shboul, M**., Lee, E., Reversade, B., Wagner, E.J., and Lee, L.A. **(2013b)**. Nuclear-localized Asunder regulates cytoplasmic dynein localization via its role in the integrator complex. Molecular biology of the cell 24, 2954-2965.

28. Jodoin, J.N., **Shboul, M**., Sitaram, P., Zein-Sabatto, H., Reversade, B., Lee, E., and Lee, L.A. **(2012)**. Human Asunder promotes dynein recruitment and centrosomal tethering to the nucleus at mitotic entry. Molecular biology of the cell 23, 4713-4724.

29. Pohler E, Mamai O, Hirst J, Zamiri M, Horn H, Nomura T, Irvine AD, Moran B, Wilson NJ, Smith FJ, Goh CS, Sandilands A, Cole C, Barton GJ, Evans AT, Shimizu H, Akiyama M, Suehiro M, Konohana I, **Shboul M**, Teissier S, Boussofara L, Denguezli M, Saad A, Gribaa M, Dopping-Hepenstal PJ, McGrath JA, Brown SJ, Goudie DR, Reversade B, Munro CS, McLean WH. **(2012)**. Haploinsufficiency for AAGAB causes clinically heterogeneous forms of punctate palmoplantar keratoderma. Nat Genet 44, 1272-1276.

30. Kroos M, Hoogeveen-Westerveld M, Michelakakis H, Pomponio R, Van der Ploeg A, Halley D, Reuser A; GAA Database Consortium. **(2012)**. Update of the pompe disease mutation database with 60 novel GAA sequence variants and additional studies on the functional effect of 34 previously reported variants. Hum Mut 33, 1161-1165.

31. Samia Temtamy, Mona Aglan, A. Kemal Topaloglu, Bernd Wollnik, Khalda Amr, Tarek H. El-Badry, Gamal A. Hosny, Nermine Salah Eldin, **Mohammad Shboul**, Mustafa Herdem, Junxian Ong, Bruno Reversade and Jing Tian. **(2012)**. Definition of the phenotypic spectrum of Temtamy Preaxial brachydactyly syndrome associated with autosomal recessive CHYS1 mutations. Middle East Journal of Medical Genetics 1, 64-70.

32. Albaramki J, Akl K, Al-Muhtaseb A, **Al-Shboul M**, Mahmoud T, El-Khateeb M, Hamamy H. **(2012)**. Sanjad Sakati syndrome: a case series from Jordan. East Mediterranean Health J 18, 527-531.

33. Dajani R, Fatahallah R, Dajani A, **Al-Shboul M**, Khader Y. **(2012)**. Prevalence of coagulation factor II G20210A and factor V G1691A Leiden polymorphisms in Chechans, a genetically isolated population in Jordan. Molecular Biology Reports 39, 9133-9138.

34. Bonnard C, Strobl AC, **Shboul M**, Lee H, Merriman B, Nelson SF, Ababneh OH, Uz E, Güran T, Kayserili H, Hamamy H, Reversade B. **(2012)**. Mutations in IRX5 impair craniofacial development and germ cell migration via SDF1. Nat Genet 44, 709-713.

35. Huang L, Szymanska K, Jensen VL, Janecke AR, Innes AM, Davis EE, Frosk P, Li C, Willer JR, Chodirker BN, Greenberg CR, McLeod DR, Bernier FP, Chudley AE, Müller T, Shboul M, Logan CV, Loucks CM, Beaulieu CL, Bowie RV, Bell SM, Adkins J, Zuniga FI, Ross KD, Wang J, Ban MR, Becker C, Nürnberg P, Douglas S, Craft CM, Akimenko MA, Hegele RA, Ober C, **Shboul M**, Utermann G, Bolz HJ, Bulman DE, Katsanis N, Blacque OE, Doherty D, Parboosingh JS, Leroux MR, Johnson CA, Boycott KM. **(2011)**. TMEM237 is mutated in individuals with a Joubert syndrome related disorder and expands the role of the TMEM family at the ciliary transition zone. Am J Hum Genet 89,713-730.

36. Tian J, Ling L, **Shboul M**, Lee H, O'Connor B, Merriman B, Nelson SF, Cool S, Ababneh OH, Al-Hadidy A, Masri A, Hamamy H, Reversade B. Loss of CHSY1, a secreted FRINGE enzyme, causes syndromic brachydactyly in humans via increased NOTCH signaling. **(2010)**. Am J Hum Genet 87, 768-778.

37. Shammas C, Papasavva T, Felekis X, Christophorou C, Roomere H, Synodinos JT, Kanavakis E, El-Khateeb M, Hamamy H, Mahmoud T, **Shboul M**, El Beshlawy A, Filon D, Hussein IR, Galanello R, Romeo G, Kleanthous M. **(2010)**. ThalassoChip, an array mutation and single

nucleotide polymorphism detection tool for the diagnosis of β-thalassaemia. Clin Chem Lab Med 48, 1713-1718.

38. Reversade B, Escande-Beillard N, Dimopoulou A, Fischer B, Chng SC, Li Y, **Shboul M**, Tham PY, Kayserili H, Al-Gazali L, Shahwan M, Brancati F, Lee H, O'Connor BD, Schmidt-von Kegler M, Merriman B, Nelson SF, Masri A, Alkazaleh F, Guerra D, Ferrari P, Nanda A, Rajab A, Markie D, Gray M, Nelson J, Grix A, Sommer A, Savarirayan R, Janecke AR, Steichen E, Sillence D, Hausser I, Budde B, Nürnberg G, Nürnberg P, Seemann P, Kunkel D, Zambruno G, Dallapiccola B, Schuelke M, Robertson S, Hamamy H, Wollnik B, Van Maldergem L, Mundlos S, Kornak U. **(2009)**. Mutations in PYCR1 cause cutis laxa with progeroid features. Nat Genet 4, 1016-21.

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