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Identification of a recognizable progressive skeletal dysplasia caused by RSPRY1 mutations

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Skeletal dysplasias are highly variable Mendelian phenotypes. Molecular diagnosis of skeletal dysplasias is complicated by their extreme clinical and genetic heterogeneity. We describe a clinically recognizable autosomal-recessive disorder in four affected siblings from a consanguineous Saudi family, comprising progressive spondyloepimetaphyseal dysplasia, short stature, facial dysmorphism, short fourth metatarsals and intellectual disability. Combined autozygome/exome analysis identified a homozygous frameshift mutation in RSPRY1 with resulting nonsense-mediated decay. Using a gene-centric matchmaking system, we were able to identify a Peruvian simplex case subject whose phenotype is strikingly similar to the original Saudi family and whose exome sequencing had revealed a likely pathogenic homozygous missense variant in the same gene. RSPRY1 encodes a hypothetical RING and SPRY domain-containing protein of unknown physiological function. However, we detect strong RSPRY1 protein localization in murine embryonic osteoblasts and periosteal cells during primary endochondral ossification, consistent with a role in bone development. This study highlights the role of gene-centric matchmaking tools to establish causal links to genes, especially for rare or previously undescribed clinical entities.

Biography

Maha Faden is an Assistant Professor at the College of Medicine at Alfaisal University, Riyadh, KSA. She is also Head of the Genetic Unit and Consultant of Pediatrics and Clinical Genetics at King Saud Medical City, Riyadh. After obtaining her medical degree from the King Abdulaziz University, Jeddah, KSA, she completed an internship year at King Abdulaziz Hospital and Oncology Centre. She was certified by the Arab Board and Saudi Board of Pediatrics in 2000. Subsequently, she achieved two fellowships – the first in clinical genetics and dysmorphology at King Faisal Specialist Hospital and Research Center, Riyadh, in 2006, and the second in skeletal dysplasia at the Cedars-Sinai Medical Center, Los Angeles, CA, USA, in 2007 as part of the University of California, Los Angeles Intercampus Medical Genetics Training Program. Since 2012, she has been involved in the newborn screening program run by the Saudi Ministry of Health and has been a regional leader for the Rare Diseases Initiative's 'Excellence in Pediatrics' conference. She is a member of several associations spanning genetics and pediatrics, has spoken at various national and international meetings, and has authored peer-reviewed publications in several international journals.

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