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Elaine Lyon
University of Utah, USA

The clinical and personal utility of multi-gene analysis for severe skeletal dysplasias

Skeletal dysplasias include over 350 disorders and have common characteristics including abnormal cartilage or bone growth with an overall incidence of approximately 1/5000. Severe skeletal dysplasias, such as thanatophoric dysplasia, osteogenesis imperfecta, achondroplasia and campomelic dysplasia can be detected prenatally by ultrasound, although correctly identifying the specific disorder is challenging. We designed and validated a multi-gene panel for sequencing genes known to be associated with severe skeletal dsyplasias. Depending on the disorder, disease prognosis varies from perinatal lethal to a normal life span. Recurrence risk for the family depends on the mode of inheritance with dominant (and presumed de novo), recessive and X-linked patterns. Because of this variability, establishing or confirming a diagnosis by molecular means has clinical and personal utility. The diagnostic efficacy (percent positive for pathogenic or likely pathogenic variants) is >50% for this prenatal skeletal dysplasia panel. Clinical scenarios illustrating the impact of accurate diagnosis, enabling treating clinicians to inform families of disease course and recurrence risk, will be presented to demonstrate the utility of this testing.

Biography

Elaine Lyon is currently a Professor of Clinical Pathology at the University of Utah, School of Medicine and a Medical Director of Molecular Genetics and Genomics ARUP Laboratories. She has received her PhD in Medical Genetics at the University of Alabama at Birmingham and completed Fellowships in Clinical Molecular Genetics and Molecular Pathology at the University of Utah. She is certified with the American Board of Medical Genetics and a Member of the Association for Molecular Pathology, American Society of Human Genetics, American College of Medical Genetics and the American Association for Clinical Chemistry.

lyone@aruplab.com