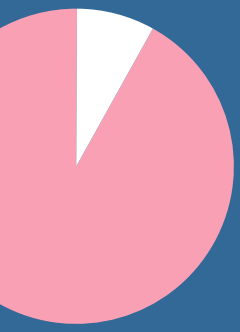
Skeletal Dysplasia



92%
of skeletal dysplasia conditions have an underlying **genetic cause**

Prevalence:
1:5,000 – 1:8,000
live births

Clinical overview

Staying up-to-date with the considerable phenotypic overlap among these conditions

The rapid advances in the field have prompted the Nosology Committee of the International Skeletal Dysplasia Society to revise and update the version of the Nosology and Classification of Genetic Skeletal Disorders.

This newest and 10th version (2019) comprises **>450** different diseases that are classified into **42** groups based on their clinical, radiographic, and/or molecular phenotypes.

https://onlinelibrary.wiley.com/doi/10.1002/ajmg.a.61366

Genetic Testing

Important that the test includes:

- ☑ Clinically relevant genes, including capabilities in difficult-to-sequence genes such as *SHOX*
- ☑ High-quality interpretation to navigate the heterogeneous genetic landscape
- ☑ High-quality sequencing performance, including for the detection of copy number variants (CNVs)
- ☑ Analysis of clinically relevant noncoding regions

When we looked at over **500** cases from **3** commonly ordered panels:

- The diagnostic yield was 42%
- CNVs accounted for more than 5% of diagnostic variants

History & Future

Short stature have been known and identified throughout recorded history, appearing in ancient Greek as well as Egyptian and Renaissance art. In the past decades, the phenotypic heterogeneity and gene discoveries have been in the focus.

Advances in research

There have been significant advances in the therapeutic development of skeletal dysplasia conditions.

For example, there are several ongoing clinical trials to improve therapeutic options for individuals with achondroplasia - the most common form of skeletal dysplasia.