Axial Spondylometaphyseal Dysplasia

**Alternative Name**
SMD, Axial
Axial SMD

**Record Category**
Disease phenotype

**WHO-ICD**
Congenital malformations, deformations and chromosomal abnormalities > Congenital malformations and deformations of the musculoskeletal system

**Incidence per 100,000 Live Births**
0-1

**OMIM Number**
602271

**Mode of Inheritance**
Autosomal recessive

**Gene Map Locus**
21q22.3 (provisional)

**Description**
Axial Spondylometaphyseal Dysplasia is an extremely rare autosomal recessive skeletal dysplasia characterized by short stature, progressive vision loss, thoracic hypoplasia, and spondylometaphyseal dysplasia. The bones of the chest, pelvis, spine, upper arms and upper legs are the ones that are primarily affected. The overall effect is one of shortened stature. Ocular anomalies seen in this condition include optic atrophy and/or retinitis pigmentosa. These features may manifest themselves in infancy or early childhood, and progress rapidly with age. Spine changes may also be seen, including scoliosis, and flattened vertebrae. Thoracic hypoplasia may lead to breathing and recurring lung infections in some cases.

The disorder is extremely rare, with less than 10 patients being reported worldwide so far. Diagnosis is achieved through skeletal survey. There is no specific treatment for the condition. Treatment is supportive, and aimed at dealing with the symptoms.

**Molecular Genetics**
Axial SMD has been noted to follow an autosomal recessive pattern of inheritance. This is reinforced by the presence of consanguinity in the affected families. The causative gene is known as C21orf2. This gene was suggested to have a role in ciliogenesis, especially pertaining to skeletal development and retinal function. It has also been speculated to play a role in the regulation of cell morphology and cytoskeletal organization.

**Epidemiology in the Arab World**

**Saudi Arabia**
Wang et al., (2016) described 13 patients from nine families with Axial Spondylometaphyseal Dysplasia (SMD). These included three patients from two Saudi Arabian families. All patients showed mild postnatal growth failure, severe thoracic deformity, retinal dystrophy and impaired visual acuity, which was noticed early in life. Radiological investigations were abnormal in all patients especially in ribs, ilia and femora. None of the patients had poliodactyly. Two of the patients from the first family suffered from cone rod dystrophy, while the third Saudi patient had retinitis pigmentosa along with photophobia. Exome sequencing identified the same novel bi-allelic mutation in C21orf2 in all three affected Saudi patients. The authors concluded that C21orf2 was the causative gene for axial SMD and that further studies were needed to clarify the role of this gene on skeletal development and retinal function.

**References**

**Related CTGA Records**
External Links
http://disorders.eyes.arizona.edu/disorders/spondylo
metaphyseal-dysplasia-axial
https://rarediseases.info.nih.gov/diseases/8720/disease

Contributor:
Ameera Balobaid: 15.10.2016