Rhizomelic Skeletal Dysplasia with Retinitis Pigmentosa

WHO International Classification of Diseases
Congenital malformations, deformations and chromosomal abnormalities

OMIM Number
609047

Mode of Inheritance
Autosomal recessive

Description
Skeletal dysplasia, rhizomelic, with retinitis pigmentosa is a very rare genetic disorder of the bones and retina described only in a Lebanese girl. The abnormalities associated with this disorder include pre- and post-natal short stature with rhizomelic limb shortening, retinitis pigmentosa, photophobia, short neck, barrel shaped chest and slight limitation of extension at the elbows. Intellectual development appears to be normal.

Molecular Genetics
It is suggested that rhizomelic skeletal dysplasia with retinitis pigmentosa is transmitted as an autosomal recessive trait since the described patient had consanguineous parents.

Epidemiology in the Arab World
Lebanon
Megarbane et al. (2004) described a girl who had a collection of abnormalities never described before. She was born to healthy first cousin Lebanese parents. The mother had two other subsequent pregnancies that ended in spontaneous abortion due to polyhydramnios and severe growth retardation. The parents had another healthy girl. When the affected girl was six months, she was diagnosed to have advanced bone age and bilateral hip dislocation. At the age of 20 months, her bone age was three years. She was first seen at 40 months of age and was noticed to have low-pitched voice and severe photophobia. Physical examination revealed low-set ears, short neck, barrel shaped chest and slight limitation of extension at the elbows. EEG showed multifocal spikes. Convergent strabismus, severe astigmatism, and flat responses in retina were identified by ophthalmological tests and electroretinogram. On radiological examination, the girl had moderate platyspondyly, ovoid vertebra, decreased interpedicular distances from upper to lower spine, abnormal ossification of the ischia, short long bones, and flared metaphyses. Her bone age was four years. Ten months later, the girl was evaluated neurologically for sub-acute episode of limping one week after an upper respiratory infection. Neurological examination and brain MRI showed significant right hemiparesis, abnormal signals on T2 in the posterior parietal cortex, centrum semi-ovale, and cortical gyrus in the left hemisphere. She had UTI with proteinuria. After two weeks, the patient recovered fully. Megarbane et al. (2004) suggested autosomal recessive inheritance for the disease due to the presence of consanguinity.

References

Contributors
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