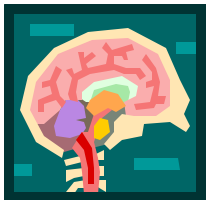

Disability Awareness Begins With You

Coffin Lowry Syndrome

Coffin-Lowry Syndrome is a rare genetic disorder characterized by craniofacial (head and facial) and skeletal abnormalities, mental retardation, short stature, and hypotonia.

Characteristic facial features may include an under-developed upper jaw bone (maxillary hypoplasia), an abnormally prominent brow, down-slanting eyelid folds (palpebral fissures), widely spaced eyes (hypertelorism), large ears, and unusually thick eyebrows.

Skeletal abnormalities may include abnormal front-to-back and side-to-side curvature of the spine (kyphoscoliosis), unusual prominence of the breastbone (pectus carinatum), and short, tapered fingers. Additional abnormalities may also be present.

Other features may include feeding and respiratory problems, developmental delay, hearing impairment, awkward gait, flat feet, and heart and kidney involvement. The disorder affects males and females in equal numbers, however, symptoms may be more severe in males.

The disorder is caused by a defective gene, which was found in 1996 on the X chromosome (Xp22.2-p22.1).

There is no cure and no standard course of treatment for Coffin-Lowry syndrome. Treatment is symptomatic and supportive, and may include physical and speech therapy.

The prognosis for individuals with Coffin-Lowry Syndrome varies depending on the severity of symptoms. Early intervention may improve the outlook for patients.

For more information, contact:

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Information from:
http://www.ninds.nih.gov/health_and_medical/disorders/coffin_lowry.htm