Coffin-Lowry Syndrome

Coffin-Lowry Syndrome

Coffin-Lowry syndrome is a genetic condition that is characterised by intellectual disability.

Individuals with Coffin-Lowry syndrome may have physical features such as large hands with short, thin fingers, short stature, small/underdeveloped upper jaw, prominent brow, and downward slanting eyelid folds.

The cause of this syndrome is a problem in the RSK2 gene, which plays an important role in the brain and its function.

Frequency

- Incidence of Coffin-Lowry syndrome is unknown, but is estimated to affect approximately 1 in 40, 000 to 50, 000 people.

Signs and Symptoms

Symptoms tend to be more severe in males. Females may have symptoms on a spectrum from mild to severe.

Facial features

- Prominent forehead and eyebrows
- Downward slanting eyelid slits
- Wide-set eyes
- Broad nasal bridge

Limb features

- Large, soft hands with tapered fingers
- Shortened big toe
- Bone abnormalities including shorter bones, or a sunken breast bone

Other features

- Short stature
- Small head
- Hearing loss
- Intellectual disability
- In rare cases vision and heart problems

Treatment

There is no specific treatment for Coffin-Lowry syndrome. Management of patients includes regular cardiac, hearing, and vision assessments

References:

Coffin-Lowry Syndrome. (2019). Retrieved from

https://rarediseases.org/rare-diseases/coffin-lowry-syndrome/

Coffin-Lowry Syndrome. (2019). Retrieved from

https://ghr.nlm.nih.gov/condition/coffin-lowry-syndrome#diagnosis

Coffin-Lowry Syndrome. (2019). Retrieved from

https://rarediseases.info.nih.gov/diseases/6123/coffin-lowry-syndrome

Coffin-Lowry Syndrome. (2019). Retrieved from https://jmg.bmj.com/content/jmedgenet/39/10/705.full.pdf

Coffin-Lowry Syndrome. (2019). Retrieved from https://clsf.info/Characteristics/Characteristics.html