# Coffin Lowry Syndrome

Coffin-Lowry syndrome is a rare genetic disorder characterized by:

- Head and Facial skeletal abnormalities.
- Mental retardation.
- Short stature.
- Hypotonia.

Facial features may include:

- An underdeveloped upper jaw bone.
- A broad nose.
- Protruding nostrils.
- An abnormally prominent brow.
- Down-slanting eyelid folds.
- Widely spaced eyes.
- Large ears.
- Unusually thick eyebrows.

Skeletal problems may include:

- Curvature of the spine.
- Prominence of the breastbone.
- Short, hyper-extensible, tapered fingers.

Other problems may also be present including:

- Feeding problems.
- Respiratory problems.
- Developmental delay.
- Mental retardation.
- Hearing impairment.
- Awkward gait.
- Flat feet.
- Heart and kidney problems.

## CAUSES

The disorder affects males and females in equal numbers. Problems may be more severe in males. Females may show mild mental retardation. The disorder is caused by a defective gene. This gene was found in 1996 on the X chromosome.

## TREATMENT

There is no cure and no standard treatment for Coffin-Lowry syndrome. Treatment is symptomatic and supportive. Treatment may include:

- Physical therapy.
- Speech therapy.
- Educational services.

## WHAT WILL HAPPEN

The likely outcome for an individual with Coffin-Lowry syndrome varies depending on the severity of symptoms. Early intervention may improve the outlook for patients.

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