



COFFIN-LOWRY SYNDROME FACT SHEET

Coffin-Lowry Syndrome (CLS) is a rare genetic disorder characterized by distinct facial features, intellectual disability, and skeletal abnormalities.

CLS was identified and named after Dr. Grange S. Coffin and Dr. Robert B. Lowry.

Genetics and Causes

CLS is caused by mutations in the RPS6KA3 gene on the X chromosome. This gene is vital for brain development.

Males exhibit more severe symptoms due to having only one X chromosome, while females may show milder symptoms or can be asymptomatic carriers. A woman who has CLS herself has a 50% chance of having a child with CLS, depending on which of her X chromosomes are inherited by the child. However, in about 70-80% of cases, the mother does not have CLS nor is there any family history – this is known as a spontaneous mutation.

Symptoms and Characteristics

Intellectual Disability

Individuals with CLS typically have developmental delays and moderate to severe intellectual disabilities.

Cranial Features

Distinctive facial features include hypertelorism, large mouth with full lips, thickened eyebrows, thickened skull, widely spaced eyes, prominent ears, early loss of teeth and missing permanent teeth.



Skeletal Abnormalities

Features can include scoliosis or kyphosis, short stature, pectus excavatum or carinatum, spinal stenosis and hyperextensible joints and delayed bone development.

Other CLS Symptoms

Additional symptoms may include seizures, cognitive/behavior issues, mobility challenges, sleep apnea, hearing loss, cardiac abnormalities, short tapered fingers.

Diagnosis

Diagnosis is based on genetic testing for RPS6KA3 mutations, clinical evaluation, physical features and family history.



Treatment and Management

Early Intervention

Early intervention with speech, occupational, and physical therapies is crucial for improving skills.

Medical Management

Medical management focuses on addressing specific symptoms and conditions, including:

- Cardiac monitoring
- Seizure management
- Orthopedic treatments
- Hearing interventions

Supportive Care

Includes educational support, social services, and genetic counseling.

Prognosis

The prognosis varies with the severity of symptoms. With appropriate care, individuals can lead fulfilling lives, although the condition may shorten life expectancy. CLS is complex and requires a multidisciplinary approach. Advances in genetic research and early interventions are improving outcomes and providing hope for affected families.



The mission of the Coffin-Lowry Syndrome Foundation (CLSF) is to provide information and support for families, caregivers, relatives, educators and medical professionals affected by, or working with, individuals diagnosed with CLS.

CLSF Vision Statement

CLSF seeks to become a visible group in the medical, scientific, educational and professional communities in order to facilitate referrals of newly diagnosed individuals, and to encourage medical and behavioral research in order to improve methods of social integration as well as treatments options for individuals with CLS.

Contact Coffin-Lowry Syndrome Foundation



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