

Inherited: SHORT Syndrome

SHORT syndrome is extremely rare and has been reported in approximately 25 patients. The SHORT syndrome has clinical manifestations of Short stature, Hyperextensibility of joints and/or inguinal hernia, Ocular depression, Reiger anomaly and Teething delay.

Reiger anomaly constitutes eye abnormalities such as hypoplasia of iris stroma, iridocorneal synechiae (adhesions between the iris and cornea), micro or megalo cornea, strabismus (stray eye), predisposition to glaucoma and tooth abnormalities like hypodontia (poor dental development), microdontia (small teeth), enamel hypoplasia (incomplete development of tooth enamel) and atypical teeth. Other clinical features may include intrauterine growth retardation with slow post natal weight gain, delayed speech with normal intellect, frequent childhood illnesses, bilateral clinodactyly (curved fingers) and sensorineuronal hearing loss (deafness), distinct facial abnormalities like disproportionately small face, sunken eyes and wide nasal bridge, inguinal hernia, heart murmur. Most of the patients have lipodystrophy involving primarily the face, upper extremities, and chest.

Distribution of fat loss is similar to that seen in patients with acquired partial lipodystrophy. Diagnosis should be suspected in patients with early onset type 2 diabetes (in 2nd or 3rd decade) who do not have obesity. Some patients can be mistakenly diagnosed as having progeria (premature aging) because of the severe lack of fat from the face. Genetic basis is unknown.

References

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