**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 2 nd or 3 rd 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**


- Sensenbrenner JA, Hussels IE, Levin LS. A low birthweight syndrome,? Rieger syndrome. Birth Defects Orig Artic Ser. 1975; 11(2): 423-6.
- Gorlin RJ, Cervenka J, Moller K, Horrobin M, Witkop J. Rieger anomaly and growth retardation (The S-H-O-R-T Syndrome). Birth Defects 1975; 11:46 -48.
- Sorge G, Ruggieri M, Polizzi A, Scuderi A, Di Pietro M. SHORT syndrome: a new case with probable autosomal dominant inheritance. Am J Med Genet 1996; 61:178-81.
- Bankier A, Keith CG, Temple IK. Absent iris stroma, narrow body build and small facial bones: a new association or variant of SHORT syndrome? Clin Dysmorphol 1995; 4:304-12.
- Aarskog D, Ose L, Pande H, Eide N. Autosomal dominant partial lipodystrophy associated with Rieger anomaly, short stature, and insulinopenic diabetes. Am J Med Gen 1983; 15:29 -38.
- Joo SH, Raygada M, Gibney S, Farzaneh I, Rennert OM. Case report on SHORT syndrome. Clin Dysmorphol. 1999 Jul;8(3):219-21.
- Bonnel S, Dureau P, LeMerrer M, Dufier JL. SHORT syndrome: a case with high hyperopia and astigmatism. Ophthalmic Genet. 2000 Dec;21(4):235-8.
- Koenig R, Brendel L, Fuchs S. SHORT syndrome. Clin Dysmorphol. 2003 Jan;12(1):45-9. Review