History
2 year old with developmental delay.

Diagnosis
Hallerman-Streiff Syndrome

Additional Clinical
History of congenital cataract resection.

Discussion
Hallermann-Streiff Syndrome (HSS) is a rare congenital anomaly that was first identified in 1893 and more completely described in 1948 by Wilhelm Hallermann and again in 1950 by Enrico Strieff. Significant craniofacial findings that characterize HSS include ocular, dental, cutaneous and skeletal abnormalities. Ocular features include microphthalmia, congenital cataracts, blue sclera, nystagmus and ptosis. In addition to ocular disturbances, 80% of children with HSS have severe dental anomalies including malocclusion, open-bite, severe caries, hypoplasia of the enamel, premature rupture of permanent teeth, neonatal teeth and hypodontia. Other classic features of HSS include dyscephaly (scaphocephaly or brachycephaly with frontal bossing), condylar aplasia, a thin pointed nose, atrophy of the skin (specifically of the head and nose), hypotrichosis, and poor development of paranasal sinuses. Cognitive function is generally spared.

Although many phenotypic characteristics have been related to HSS, seven essential signs are commonly used to define this anomaly. These essential findings are: 1) dyscephalia and bird-like facies, 2) abnormal dentition, 3) hypotrichosis, 4) atrophy of skin, especially on the nose, 5) congenital cataracts, 6) bilateral microphthalmia, and 7) proportionate dwarfism. Oculodentodigital dysplasia, progeria, Bloom syndrome, Rothmund-Thompson syndrome, Seckel syndrome have phenotypic similarities but no more than two of the seven essential signs.

Most documented cases of HSS are sporadic; however, cases of autosomal dominant and recessive inheritance patterns have been reported. The potential causes of HSS include: a brachial arch defect, exposure to toxins, maternal viral infections and advanced paternal age. A diagnosis of HSS carries a poor prognosis. Death generally occurs during infancy from respiratory difficulties related to small nares and glossoptosis, which are secondary to micrognathia and tracheomalacia. Treatment is directed toward airway management, vision correction, craniofacial reconstruction and dental care.

Findings
MR -1) Microophthalmia with resected lenses and 2) Micrognathia.

Reference

**Contributor**
Cole Denton
Orrie Close
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