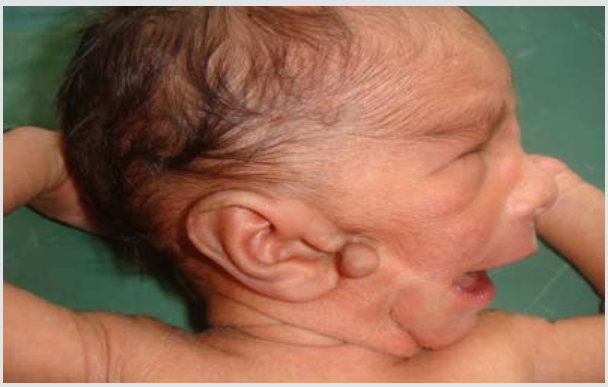
SPOT DIAGNOSIS (IMAGE GALLERY)



FACIAL TAGS -GOLDENHAR SYNDROME

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Spot Diagnosis

Goldenhar syndrome. This child has hemifacial microsomia with evident fusion line of maxillary and mandibular processes starting from tragus up to angle of mouth on right side. There is philtrum and thin upper lip with low set ears, with bilateral multiple pre-auricular tags on right side. Goldenhar syndrome was first described by Dr. Maurice Goldenhar in 1952. Goldenhar syndrome occurs in one of every 3,000 to 5,000 live births. Males are affected more frequently than females. The abnormalities seen in Goldenhar syndrome are typically limited to the face and vertebrae. (1) Thirty percent of patients have bilateral facial abnormalities. In these patients, the right side is usually affected more severely. The symptoms associated with Goldenhar syndrome are highly variable. Some individuals with Goldenhar syndrome have many severe abnormalities, while other individuals have few minor birth defects. Hemifacial microsomia is a common physical difference seen in Goldenhar syndrome. (2) This is caused by hypoplasia of the bones of the face mainly the mandible and the maxilla. In addition to the bones of the face, the muscles of the face can also be underdeveloped. Cleft lip and cleft palate are another facial difference associated with Goldenhar syndrome. Macrostomia is often present. Abnormal development of the ears is another characteristic of Goldenhar syndrome. The ears may be smaller than normal (microtia), or absent (anotia). Ear tags (excess pieces of skin) may be seen on the cheek next to the ear and may extend to the corner of the mouth. The shape of the ears may also be unusual. Hearing loss is common in individuals with Goldenhar syndrome. (1) The vertebral problems seen in Goldenhar syndrome result from incomplete development of the vertebrae. Vertebrae can be incompletely developed (hemivertebrae), absent, or fused. Ribs can also be abnormal. Approximately 15 percent of individuals with Goldenhar syndrome have developmental delay or mental retardation. (2)

References:

1. Michael CM, Gorlin R, Fraser FC. Craniofacial Disorders. In Emery and Rimoin`s Principles and Practice of Medical Genetics. 3rd ed. New York, Churchill Livingstone. 1996: 1132-1134
2. Jones KL. Oculo-auriculo-vertebral spectrum (First and second branchial arch syndrome, facio-auriculo-vertebral spectrum, hemifacial microsomia, Goldenhar syndrome). In: Smith`s recognizable patterns of human malformation, 6th ed, Elsevier Saunders, Philadelphia 2006. p.738.

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