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# Goldenhar syndrome and pericentric inversion of chromosome 9

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## Abstract

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Oculo-auriculovertebral dysplasia (Goldenhar) is a congenital syndrome. Its phenotype differs from craniofacial anomalies to cardiac, vertebral or central nervous system defects. This syndrome is rare and its etiology is not apparent yet. Pericentric inversion of chromosome 9 is one of the most common structural balanced chromosomal aberrations with its incidences 15% to 25%. Herein we present a case of Goldenhar syndrome in a one-yr-old girl with pericentric inversion of chromosome 9. We used the patient's peripheral blood and studied 30 metaphase spreads on the basis of G-bands by trypsin using Giemsa (GTG) technique at 400 band resolution that revealed a pericentric inversion of chromosome 9 with break points at p11 and q13.

## Reaxys Database Information

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## Author keywords

Chromosome 9; Goldenhar syndrome; Pericentric inversion

## Indexed Keywords

EMTREE drug terms: trypsin

EMTREE medical terms: article; case report; chromosome 11p; chromosome 13q; chromosome analysis; chromosome breakage; chromosome inversion 9; clinical examination; clinical feature; computer assisted tomography; electroencephalogram; female; Giemsa stain; Goldenhar syndrome; human; infant; metaphase; neurologic examination; panoramic radiography; pericentric chromosome inversion

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