

## A case of Goldenhar syndrome

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### Objective

The objective of this paper is to describe a case report of patient with prenatal diagnosis de Goldenhar syndrome. Suspected by facial dysmorphias and congenital heart disease.

### Methods

Review of ultrasound images, data and medical file of patient of CIMAF (Center of Maternal Fetal Investigation) Sotero del Rio's Hospital, Santiago, Chile.

### Results

We describe the case of a 32 year old patient who was referred at 28 weeks' of pregnancy to our center for suspicions of congenital heart disease. Ultrasound findings were: hypoplastic nasal bone, prenasal edema, micrognathia, short implantation ears, bilateral abnormalities in the external ears. Cardiac evaluation showed interventricular septal defect (IVSD) and Truncus arteriosus type 1. Prenatal studies: amniocentesis for fetal karyotype (results 46, XY) Serial follow ups in our unit, normal growth (25<sup>th</sup> centile for local growth curves). The fetus was delivered by cesarean section (due to previous cesarean section) at 39<sup>+6</sup> weeks. Fetal outcome: Weight 3160 grams, 49cm, Apgar 6-9, hypotonic, respiratory failure requiring mechanical ventilation for a few days. Postnatal evaluation by the genetics team confirmed Goldenhar syndrome. Cardiac surgery was performed at 10 days for correction of the truncus arteriosus and IVSD. Due to difficult airway management a tracheostomy was performed after cardiac surgery. The newborn remained an inpatient at time of this report. Array comparative genomic hybridation (CGH) is pending.

### Conclusion

Goldenhar syndrome is a rare genetic syndrome involving many facial and cardiac defects. Prenatal diagnosis is important due to difficult management of the airway, and the need for cardiac surgery. Multidisciplinary team work is needed, as well as to deliver at a tertiary center.

