Case reports

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Tracheo-oesophageal anomalies in the Goldenhar anomalad

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SUMMARY A case of the Goldenhar anomalad is presented with a previously undescribed association with oesophageal atresia and tracheo-oesophageal fistula. This is the second instance of a tracheobronchial-oesophageal communication being found in association with the anomalad. Awareness of this combination may facilitate future diagnosis and treatment of the anomaly.

The Goldenhar anomalad is an association of facial and auricular anomalies resulting from errors in morphogenesis of the first and second branchial arches, accompanied sometimes by vertebral, ocular, and visceral malformations.1 We describe a case with previously undescribed anomalies including tracheo-oesophageal fistula, oesophageal atresia, and hypoplasia of the cerebellum.

Case report

A 2-025 kg female infant was delivered after 36 weeks of uneventful gestation to a 24 year old mother. The parents were unrelated Sephardic Jews and both they and their two children were normal and in good health. The Apgar scores were 6 and 9 at 1 and 5 minutes respectively. Increasing dyspnoea and excessive salivation were soon noted and an attempt to pass an orogastric catheter was unsuccessful.

On examination the baby was noted to have an asymmetrical facies with maxillomandibular hypoplasia, bilateral preauricular skin tags, and microphthalmia with lipodermoids on both lower palpebrae (figure a). The cardiac impulse was felt over the right chest while breath sounds were heard over the left side only. The rest of the examination was considered normal. On chest x-ray a hyperinflated left lung with a complete rightward shift of the mediastinal structures and opacification of the right chest was seen. Extranumerary ribs and hemivertebrae of D1 to D4 were present. The visible tip of the orogastric catheter at D3 and the presence of gastric air suggested the presence of oesophageal atresia with a tracheo-oesophageal fistula. Bronchography showed a complete absence of the right bronchial tree (figure b). The baby died at 20 hours of age.

At necropsy the right lung and the right mainstem bronchus were absent. The left lung had three lobes. The upper portion of the oesophagus was separated from the lower half and ended blindly. The lower portion opened into the trachea. The cardiac chambers and valves were normal. The ductus arteriosus and the foramen ovale were patent. The pulmonary trunk divided into two branches, both supplying the

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left lung. There were only two pulmonary veins draining the left lung. A small spleniculus was found at the hilus of the spleen. The cerebellum was markedly hypoplastic.

Discussion

Since the first description of the Goldenhar anomalad in 1952,2 variable combinations of facial, oral, auricular, vertebral, ocular, and cardiac malformations have been described. Other occasional abnormalities include branchial cleft remnants, laryngeal anomalies, hypoplasia or aplasia of the lung, occipital encephalocele, and renal and limb anomalies.1 The combination of oesophageal atresia and pulmonary agenesis has been reported in 14 cases, recently summarised.3 Such an association with the Goldenhar anomalad has not been previously described. Bowen and Parry,4 however, described a patient with bronchopulmonary-foregut communication and hypoplasia of the right lung but with a patent oesophagus. Thus, our report is the second instance where tracheobronchial-oesophageal communication was found in association with the Goldenhar anomalad. Awareness of this combination may facilitate future diagnosis and treatment of the anomaly.

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Severe pseudoachondroplasia with parental consanguinity

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SUMMARY A boy who showed features of the severe form of pseudoachondroplasia, whose parents were first cousins, is reported. Published reports supporting the existence of an autosomal recessive form of this disorder are reviewed.

Pseudoachondroplasia comprises a relatively common form of short limbed dwarfism with involvement of the spine, metaphyses, and epiphyses, but not the skull. Clinically different grades of severity can be recognised and provisionally four forms, severe and mild, dominant and recessive, have been identified.1 In this report a child who showed features of severe pseudoachondroplasia is described. The parents were first cousins.
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