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Aplasia of the parotid gland in Down syndrome

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Summary Salivary gland aplasia has not to our knowledge been previously reported in association with Down syndrome. We present a case of bilateral parotid aplasia in a patient with Down syndrome. Clinically he had aplasia of the major salivary glands and symptoms of xerostomia. Thirteen other family members over three generations were examined, and all had functional parotid glands. We reviewed publications about Down syndrome and salivary aplasia, together with the data regarding his other clinical problems and family background. His oral problems were inadequate plaque control, dental caries, and erosion of the teeth.

Introduction

The worldwide incidence of Down syndrome at birth is about 1/1000 live births, the incidence rising with increasing maternal age. About 95% of patients with Down syndrome, have trisomy 21. A further 4% have translocation between all or part of chromosome 21 and another of the acrocentric chromosomes, usually chromosome 14. The remainder have mosaicism, with some cell lines carrying a normal set of chromosomes while others have trisomy 21.

Gastrointestinal disorders are common in patients with Down syndrome. Atresia, tracheo-oesophageal fistula, gastro-oesophageal reflux, hiatus hernia, and non-specific motility disorders are common abnormalities. Acid reflux is more common in patients with Down syndrome and up to 10% of the children who have hiatus hernias repaired have been reported to have Down syndrome. Achalasia is an uncommon oesophageal motor disorder characterised by lack of peristaltic contractions of the oesophageal body, increased lower oesophageal sphincter pressure, and impaired relaxation during swallowing. It has been suggested that Down syndrome might be associated with a higher incidence of achalasia.

Hearing loss is common in Down syndrome, and sensorineural and mixed losses may be evident. Histopathological findings in the temporal bone include hypoplasia, sclerosis, and residual mesenchyme in the middle ear cleft. The high incidence of conductive hearing loss may be attributed to dysfunction of the Eustachian tube that leads to chronic otitis media and abnormalities of the ossicular chain—usually of the stapes. Narrow auditory canals are also common. There may be an underlying sensorineural defect, a short cochlea being the most common finding.

Older children and adults with Down syndrome have an increased incidence of autoimmune disorders affecting thyroid glands, pancreas, gastric mucosa, and adrenal glands. Thyroid autoantibodies are often found: both hyperthyroidism and hypothyroidism are common.

The facial appearance alone is often enough to make a clinical diagnosis. The cranium is brachycephalic, with underdevelopment or hypoplasia of

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the midfacial region. The bridge of the nose and the maxilla are relatively smaller. Lack of or reduction in size of, the frontal and maxillary sinuses is common. Patients with Down syndrome usually have a narrow and high vault. The obtuse nasion—sella—basion (NSB) angle leads to a flat cranial base. While the midface tends to be more deficient than the mandible, the mandibular size is also mildly hypoplastic by the early teens.

Reduced muscle tone affects the perioral muscles, which leads to a descending angle of the mouth, a raise of the upper lip and everted lower lip with protrusion of the tongue. The tongue and lips often develop fissures and the lateral border may show tooth indentations that result from hypotonia of the lingual musculature.

Defective development of the teeth is 10 times more likely in people with Down syndrome than in the general population. The teeth most often affected by agenesis are the central incisors, followed by maxillary lateral incisors, second premolars, and mandibular second premolars. Dental characteristics include delayed eruption, microdontia, hypoplasia, and taurodontism. Mottled teeth and teeth with thin enamel are also common. A study showed that supernumerary teeth were seen in 6% of 70 patients with Down syndrome.[1] There was coexisting hypodontia and supernumerary teeth in two patients. Most patients with Down syndrome have occlusal problems. Hypotonia of the orofacial muscle tissue and tongue can also cause an imbalance of the forces, contributing to an open bite.

Case report

A 24-year-old man with Down syndrome presented with a persistent dry mouth. He was white and born when his mother was 32 years old. He was 153 cm tall and weighed 43 kg. Chromosome analysis showed a karyotype of 47; XY, +21.

His elder sister was healthy and she, together with 13 other family members spanning three generations, were examined. All were found to have functional parotid glands although three had either a high incidence of caries or dry mouth.

Continuous speech was difficult because of lack of saliva and dysphagia. He was able to ingest only homogenised foods and had intermittent episodes of respiratory distress that was attributed to gastro-oesophageal reflux. This is currently under review. In infancy he had bilateral inguinal hernias. There were no symptoms to suggest a connective tissue disorder or any other systemic illness. His hearing was impaired but the nature of the defect is unknown.

He seemed healthy in general and his hair, nails, and sweat glands were all within normal limits. On physical examination no masses were palpated in the cervical region and there was a depression between the posterior border of the mandible and the mastoid process. The lips were dry and fissured. Intraorally the mucosa was dry and his tongue was fissured. There were no parotid duct orifices, but clear saliva was seen flowing from the submandibular ducts.

His oral hygiene was poor despite visits to his own private dentist for dental hygiene every 6 weeks. There was plaque throughout the gums bled when probed, and there was marginal gingivitis. He had much caries. The palatal surfaces of the upper anterior teeth and lingual surfaces of the lower molars were eroded. The palatal vault was high and the mucosa red: candidal hyphae were present on a palatal smear.

Investigations

The complete blood count and erythrocyte sedimentation rate (ESR) were within reference ranges, as were fasting plasma glucose and glycated haemoglobin concentrations. Serum proteins, liver function tests, immunoglobulins and electrolytes were also within reference ranges. Anti nuclear factor, rheumatoid factor, anti-Ro and anti-La were not recorded. There was no reaction to thyroglobulin antibodies but there was to thyroid microsomal antibodies at a titre of 1:1600. The concentrations of T3 and T4 were with reference ranges, but the concentration of thyroid stimulating hormone (TSH) was marginally raised. This was consistent with early hypothyroidism.

A ^{99m}Tc-pertechnetate scan showed uptake within the normal range in the submandibular region as well as over the thyroid, but there was no uptake in the parotid regions on either side.

Discussion

The major salivary glands begin as solid proliferations of cells from the oral epithelium between 6 and 8 weeks in utero. This epithelial mass branches into the underlying mesenchyme to form cords, the terminus of each branch being a rounded, radial array of cells. Later, as these cords become canalised, they form ducts, and the terminal cells differentiate into acini. The surrounding mesenchyme becomes condensed to form the stroma and fibrous capsule. The parotid glands develop from ectoderm while the submandibular and sublingual