Dental findings associated with the malformations of CHARGE

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Abstract

The acronym CHARGE refers to a non-random clustering of congenital malformations whose cause remains unknown. To date, the dental features of CHARGE association are not well known. A brief review of the literature and a case in a 10-year old boy with the CHARGE association are presented. The patient had multiple dental anomalies including congenital absence of teeth, ectopic eruption, submergence of primary molars and an odontogenic fibroma associated with an impacted permanent molar. *(Pediatr Dent 24:43-46, 2002)*

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In 1979, a group of congenital anomalies was identified by Hall.[1] This association of characteristics was given the acronym of CHARGE, representing an anomaly consisting of the following:[2]

C - coloboma associated with the choroids, iris, retina or optic nerve;
H - heart disease, which may include tetralogy of Fallot, septal defect, or valvular stenosis;
A - atresia of choanae;
R - retarded mental development (developmental delay);
G - genital hypoplasia;
E - ear anomalies and /or deafness.

A range of additional congenital abnormalities is now accepted to form part of the association, notably unilateral facial nerve palsy, orofacial clefts, renal abnormalities and characteristic facies. The latter has been described as a square face, with malar flattening, pinched nostrils and a prominent columella.[3] Originally, the diagnosis of CHARGE association required that at least four of the six characteristics be present.[4] Major, intermediate and minor criteria were proposed by Mitchell *et al.*[5] Recently, Blake reviewed the clinical characteristics and management of the CHARGE association.[4]

Major criteria are those findings that occur commonly in the CHARGE association but are relatively rare in other conditions: coloboma, choanal atresia, cranial nerve involvement (often multiple) and characteristic ear abnormalities. Of these, choanal atresia can have significant impact perinatally. Bilateral choanal atresia often results in breathing difficulties and cyanosis in the first hour of life. Crying may mask the cyanosis as it forces the obligate neonatal nose breather to take air by mouth. The cyanosis then returns when the infant quiets. Feeding may also be difficult, exacerbating symptoms of cyanosis.[4] An oral airway (obturator) can help relieve symptoms. Downward pressure on the mandible and/or pulling the tongue down can help give temporary relief.[4] Ultimately, surgical opening of the posterior choanae is necessary.

Bilateral stenosis and unilateral choanal atresia often go unrecognized in the neonatal period although, even in these cases, surgery is often necessary due to breathing difficulties.[4]

The minor characteristics occur less frequently, or are less specific to CHARGE: heart defects, genital hypoplasia, orofacial clefting, tracheoesophageal fistula, short stature and developmental delay. Occasional less specific findings include: renal anomalies, thymic/parathyroid hypoplasia, hand and spine anomalies, webbed neck, sloping shoulders, nipple anomalies, umbilical hernia and difficulty in swallowing.

The etiology of the CHARGE association remains unknown. Although autosomal dominant and recessive inheritances are suggested by occurrence in some families, this association is mostly reported in sporadic cases. It is believed to occur in 1:10,000 live births.[6] No teratogens have been implicated so far in the etiology of the CHARGE association, so accurate periconceptional and prenatal histories are necessary in future research for the discovery of the cause or causes of the association.

Dental findings have been reported in a few publications; Venetikidou[7] in 1993 reported two patients who exhibited

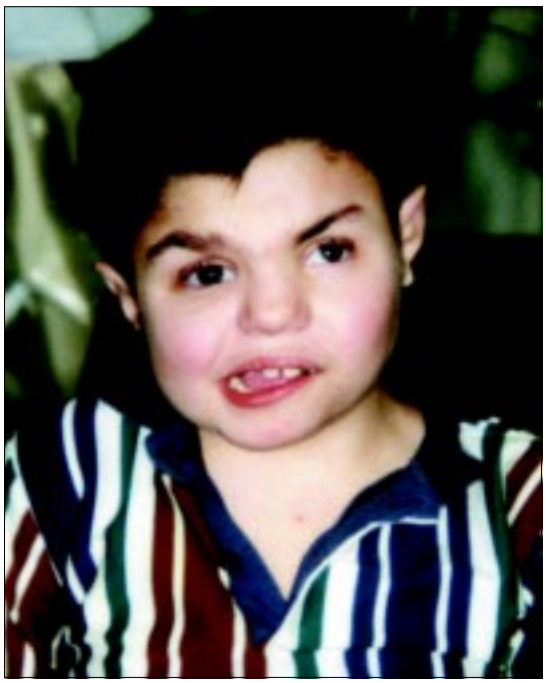

Fig 1. Facial asymmetry and left optic nerve coloboma

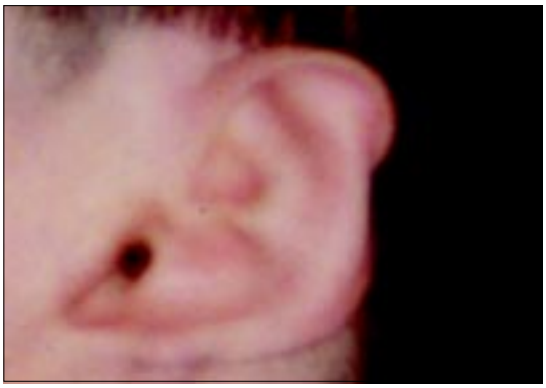

Fig 2. Ear abnormalities

delayed eruption of the permanent dentition and mandibular retrognathism. An absence of lower permanent central incisors was also described in one case. Harrison *et al* [8] reported two cases of solitary maxillary central incisor in children with CHARGE association. Grimm *et al* [9] reported taurodontism of the pulp chambers of the primary molars in a child with CHARGE association. This report describes the dental clinical findings in a child with CHARGE association.

Case report

A 10-year-old male with CHARGE association was referred by his general medical practitioner to the Pediatric Dentistry Clinic at the Franciscan Children's Hospital, Boston, MA. The chief complaint of the mother was "He has horrible teeth and needs a dentist who treats patients with special needs."

Medical history

The boy was diagnosed with CHARGE association at 13 weeks of age and is followed extensively at Children's Hospital, Boston. He was born at about 36 weeks of gestation by Cesarian section. There were no complications during pregnancy, and no particular medication was administered during pregnancy. The immediate perinatal and antenatal period was uneventful. The family includes healthy parents with normal chromosomes. He has twin siblings (brother and sister) and a younger brother. All siblings are unaffected and do not show any features of CHARGE association.

Unilateral choanal atresia is present. Cardiac abnormalities include a small atrial septal defect and a cleft mitral valve with mild mitral regurgitation. Future heart surgery will be necessary and he requires Subacute Bacterial Endocarditis prophylaxis for dental treatment. He has a left optic nerve coloboma. His ears show abnormalities of the small lobule and the lower helical fold is thin. He has bilateral PE- tubes.

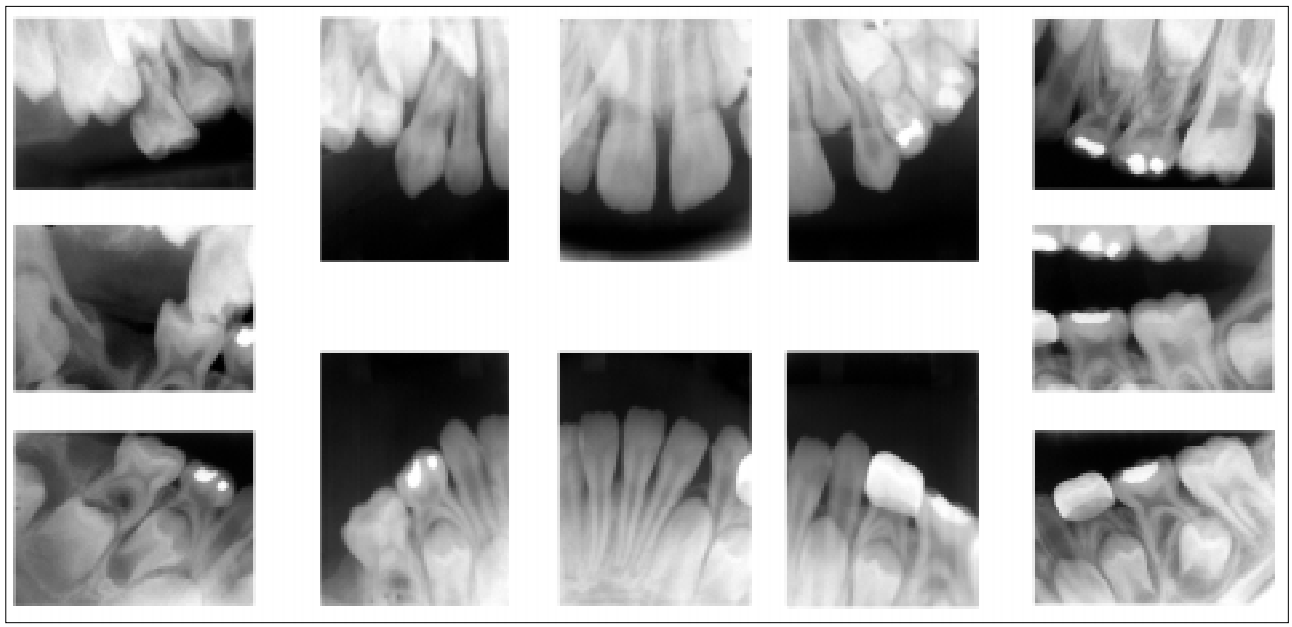

Fig 3. Full mouth radiographs at first visit

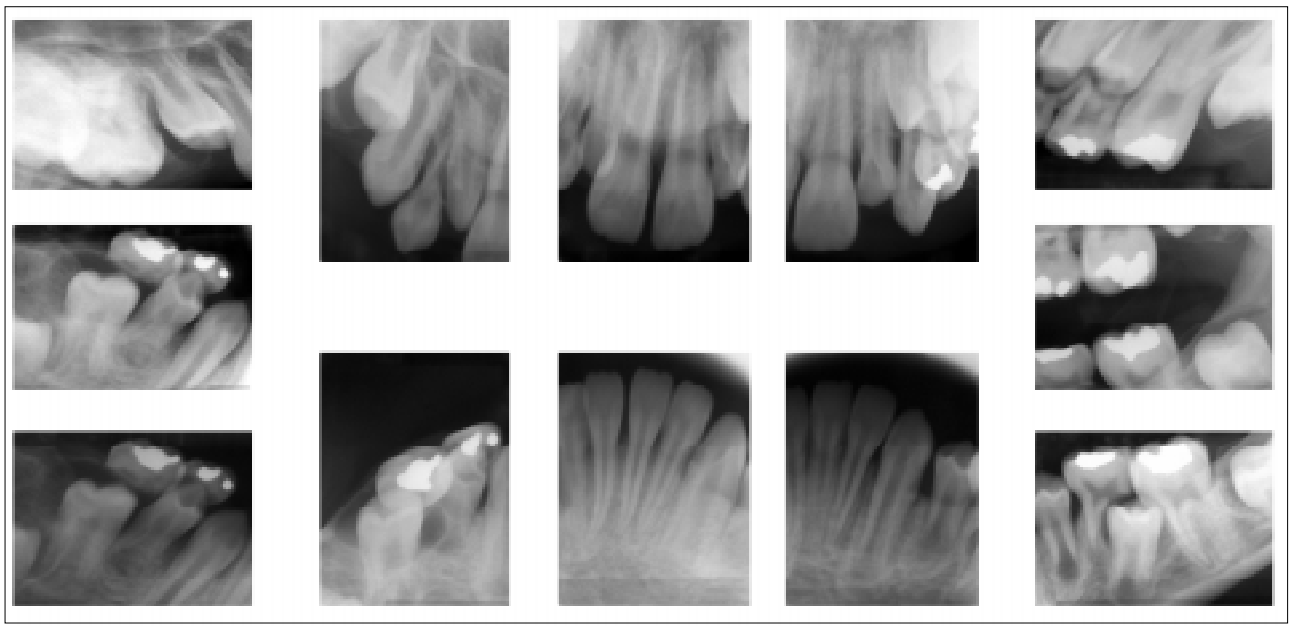

Fig 4. Full mouth radiographs one year later

He has a short neck with possible Sprengel's deformity and scoliosis. The patient demonstrates a severe oro-motor aversion to food and liquids and is entirely tube fed with a gastrostomy tube. He has a low normal IQ. He is able to ambulate and run, but has a tight heel cord and walks with a distorted gait. At age 8, his height was 48 inches, which puts him in the 25th percentile of his age, and his weight was 72 pounds, which places him in the 90th percentile.

Dental history and findings

According to the patient's mother, the boy had oral rehabilitation under general anesthesia (GA) in 1996 at another facility. Since that time, his dental care had been sporadic. During his initial evaluation in our clinic, a full medical and dental history was obtained. On extraoral examination, the patient had pronounced facial asymmetry with a convex facial profile and left optic nerve coloboma (Fig 1). The ears had small lobules, and bilateral lower helical folds were thin (Fig 2). On intra-oral examination, the dental findings included moderate to severe gingivitis and very poor oral hygiene with high levels of plaque, calculus and staining. Numerous carious primary and permanent teeth were also noted. Other clinical findings included high maxillary frenum attachment and constriction of the palate. The mother also reports mouth breathing and self-stimulatory behaviors including biting hands and gnashing teeth.

Due to the patient's medical history and behavioral difficulties encountered during the first visit, consent was obtained from the parents for further radiographic evaluation and treatment under general anesthesia. At that time, a full mouth radiographic series was obtained (Fig 3) and a dental treatment plan was developed. Multiple dental anomalies were noted in the radiographic examination completed at that time including:

1. Ectopic eruption of tooth #3 (the maxillary right first permanent molar).

Fig 5. Unerupted maxillary right permanent molar

2. Submergence of tooth #B (the maxillary right first primary molar).
3. Congenital absence of tooth #5 (the maxillary right second premolar).
4. Congenital absence of tooth #11(the maxillary left permanent canine).
5. Impaction/displacement of teeth #30 and #31 (the madibular right first and second permanent molars) with associated radiolucency.

Due to the presence of multiple dental anomalies, orthodontic and oral surgical consultation was sought. However, the patient did not present for initial follow-up and returned for recall evaluation six months following the operating room (OR) visit. At that time, oral surgical consultation was obtained and the decision made to return to the OR for surgical exploration and treatment of teeth #31 and #30 and the associated radiolucency. The surgical event was undertaken almost a year after the first OR visit. A full mouth radiographic series was obtained (Fig 4) confirming the dental anomalies which were noted in the previous radiographs.

At this time, an unerupted maxillary right first permanent molar (Fig 5) and impacted mandibular right first permanent molar at the age of 10 years indicated the delayed eruption of certain permanent teeth. After exposing part of the impacted tooth, a suture needle was placed in that region and a periapical radiograph was taken to confirm the surgical location (Fig 4).

Due to the high risk of jaw fracture and injury to the alveolar nerve and after discussion with both parents, it was decided to remove tooth #31, which was malpositioned, mesially angled and buccally positioned within the bone. Surgical curettage of the radiolucent lesion was also performed and tissue submitted for pathological examination. Mandibular right first and second primary molars as well as the mandibular left second primary molars were also removed due to mobility. The surgical pathology report indicated the presence of fragments of fibrous connective tissue with mucinous degenerative changes associated with minute fragments of bone and epithelium consistent with an odontogenic fibroma. Routine dental and preventive procedures were also completed. The postoperative course was uneventful. The patient was seen for follow-up two weeks later with no complications and good healing.

Discussion

In this report, a rare case of CHARGE association with unusual dental findings is presented. The patient presents with choanal atresia, congenital heart disease expressed as an atrial septal defect, developmental delays, neurological abnormalities and dysmorphic facial features that are in agreement with the description of the association.[9]

Although many reports of the CHARGE association appear in the literature, only a very limited number of these report dental findings. In the current case, a number of dental findings, some previously unreported in CHARGE association patients, are seen. Delayed eruption of the permanent teeth, previously reported by Venetikidou (1993) is seen in the current case. Harrison *et al*[8] reported congenitally missing central incisors in two CHARGE association patients; likewise our patient is congenitally missing the maxillary first premolar and the contra lateral maxillary canine. Congenital absence of maxillary canines is decidedly rare with only a few cases reported in the literature.[10]

Of particular interest are the impaction of tooth #30 and its association with a radiolucent lesion. It appears the lesion had led to displacement of tooth #31. Impaction of tooth #30 and ectopic eruption of tooth #3 may be related to the right side hypoplasia and obvious facial asymmetry observed in the patient. Choanal atresia, maxillary hypoplasia and hypodontia have recently been reported together in an extended family.[10] Pathological examination of tissues obtained at biopsy from the lesion suggests the diagnosis of odontogenic fibroma. These types of lesions are very unusual in children and there are no previous reports of their occurrence in CHARGE association.

Finally, ectopic eruption of maxillary permanent first molars and submergence of primary first molars, while relatively common events, have not been previously reported in CHARGE association patients. Certainly, clinical findings in this patient and other case reports suggest that careful radiographic monitoring of CHARGE association patients is justified, particularly in those patients exhibiting pronounced facial asymmetries.

Previous dental findings in CHARGE association patients have included congenital absence of maxillary central incisors and delayed eruption of permanent teeth. The current case, a 10-year-old boy with CHARGE association, exhibits multiple dental anomalies including congenital absence of teeth, ectopic eruption of maxillary permanent first molars, submergence of primary molars and an odontogenic fibroma associated with impacted mandibular first permanent molar. Careful clinical and radiographic monitoring should be part of the routine care of CHARGE association patients.

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