Blepharo-chelio-odontic Syndrome - A Case Report

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Introduction

Blepharo-cheilo-odontic syndrome (BCD syndrome) is an autosomal dominant genetic condition. Fewer than 1,000 people in the U.S. have BCD syndrome and symptoms may start to appear as a newborn. BCD syndrome mainly affects the eyelids, upper lip, and teeth. The eyelid anomalies distinguish BCD syndrome from other syndromes that also manifest with features of cleft lip and palate with oligodontia. This report highlights the assessment, diagnosis, and treatment planning considerations for a 14 year 5 month female patient with type 2 BCD syndrome.

Etiology & Genetics

BCD syndrome is due to either heterozygous CDH1 (type 1) or CTNND1 (type 2) mutations. BCD related to CDH1 and CTNND1 variants are clinically indistinguishable, although CTNND1 usually shows a milder phenotype. CDH1 encodes the epithelial cadherin, a transmembrane glycoprotein. CDH1 gene mutations may result in an E-cadherin protein that is unstable and quickly broken down. CTNND1 encodes the delta-catenin, a protein linked to the cytoplasmic domain of E-cadherin. CTNND1 gene mutations reduce or eliminate the production or function of p120-catenin, so E-cadherin is broken down prematurely. Both genes code for proteins that are involved in a cell-to-cell adhesion complex that is critical for establishing and maintaining polarized and differentiated epithelia during development. Interactions between the two proteins are also important for other cell processes that are involved in craniofacial development.

Diagnosis

BCD syndrome is suspected with the association of CLP, eyelid manifestations, and ectodermal dysplasia features. Diagnosis involves molecular genetic testing, targeting sequencing of the entire CDH1 and CTNND1 coding regions. Prenatal testing is possible if the pathogenic variant has been identified in an affected family member. If the pathogenic variant is identified in the fetus, the severity of the condition is not predictable.

Clinical Manifestations

Eye Manifestations

-Abnormalities of vision

-Distichiasis: double rows of eyelashes

-Ectropion of lower eyelid: lower eyelid turned out

-Euryblepharon: lower lid sags away from they eye

-Lagophthalmia: eyelids not able to close completely

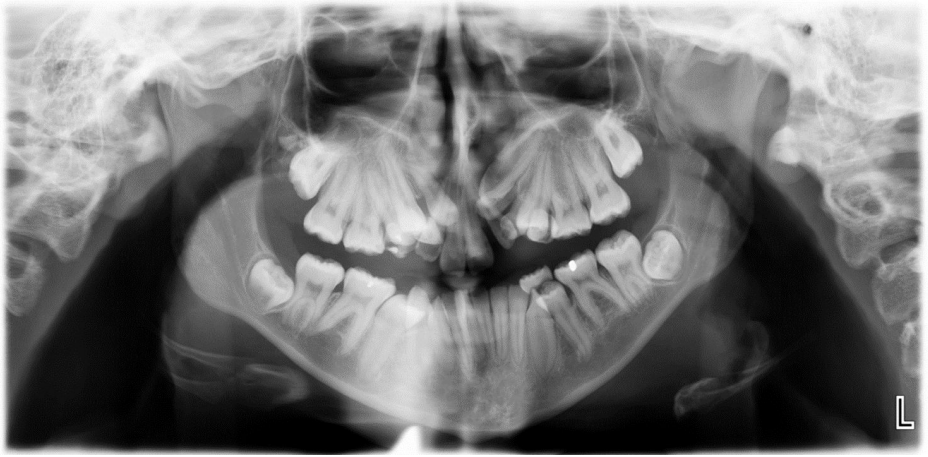
-Hypertelorism: increased interpupillary distance

Craniofacial Manifestations

-Bilateral cleft lip and palate

-Oligodontia

-Conical teeth



Clinical Management

Treatment of BCD syndrome is based on treating the symptoms and signs. A multidisciplinary approach for evaluation and treatment encourages collaboration between ophthalmologists, optometrists, endocrinologists,and pediatricians. Genetic counseling is recommended for the family autosomal dominant inheritance pattern.

The dental management team should include a craniofacial team with pediatric dentists, orthodontists, and oral and maxillofacial surgeons. The dental abnormalities affect both primary and permanent teeth. The pattern of tooth agenesis/oligodontia is unusual, involving teeth that are normally more resistant to agenesis, such as central incisors and canines. As the patient ages multi-speciality collaboration is critical for treatment of missing teeth.

Case Report (14y5m F)

Health History: CTNND1 related BCD syndrome, autism, bilateral cleft lip and palate, Marcus Gunn jaw winking, obstructive sleep apnea, hyperopia with astigmatism, bilateral exotropia, hyponasality, eustachian tube dysfunction, cerumen impaction, innocent heart murmur, and pronation of foot

Medications: loratadine (Claritin), diphenhydramine (Benadryl)
Allergies: NKDA, bee pollen (edema)

BCD Syndrome History: She has a truncated mutation of CTNND1 that has not been reported in the literature thus far. The CTNND1 mutation is attributed to BCD syndrome and thought to be an explanation for her cleft lip and palate, dental anomalies (missing and supernumerary teeth), and visual anomalies.

Surgical History: cleft lip and palate repair, bilateral tympanostomy tube placement, right eye muscle correction, tonsillectomy and adenoidectomy

Clinical exam: OH is poor, caries risk is high, mixed dentition, bilateral class III molar relation, OB: 80%, OJ:-2mm, full arch crossbite, midline deviation, maxillary and mandibular crowding

Clinical caries: #2-O, #14-O, #18-O
Other findings: congenitally missing #20 and #29, supernumerary mandibular incisor

Referred from the Hospital Dentistry Clinic at C.S. Mott Children's Hospital to the UofM Pediatric Dental Clinic for restorations and extraction of #B, #C, #H, #J, & #K due to ectopic eruption

Citations

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