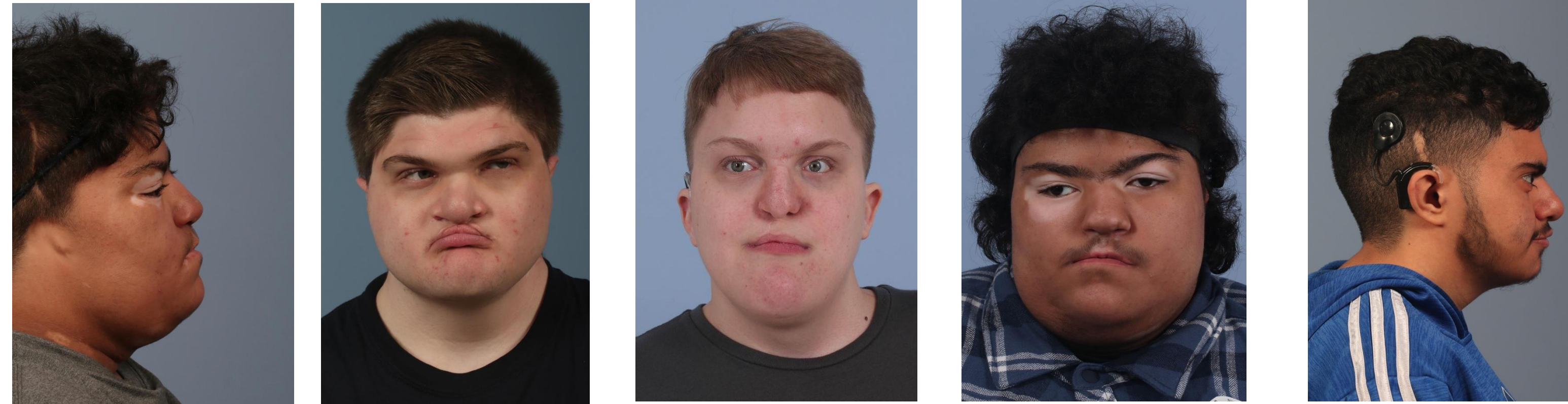




## OVERVIEW

CHARGE syndrome is a complex genetic disorder most commonly caused by a new mutation in the *CHD7* gene, though autosomal dominant inheritance can occur. Affecting approximately 1 in 8,500 to 10,000 newborns, it is often identified prenatally or in the neonatal period due to the presence of multiple congenital anomalies. The condition is characterized by a constellation of features summarized by the CHARGE acronym: coloboma and cranial nerve abnormalities, heart defects, atresia of the choanae, restriction of growth and development, genital abnormalities, and ear anomalies. Beyond these hallmark findings, many individuals also experience feeding and swallowing difficulties, cleft lip or palate, hypotonia, and kidney abnormalities, contributing to the significant medical complexity associated with this syndrome.

## CLINICAL PHOTOS

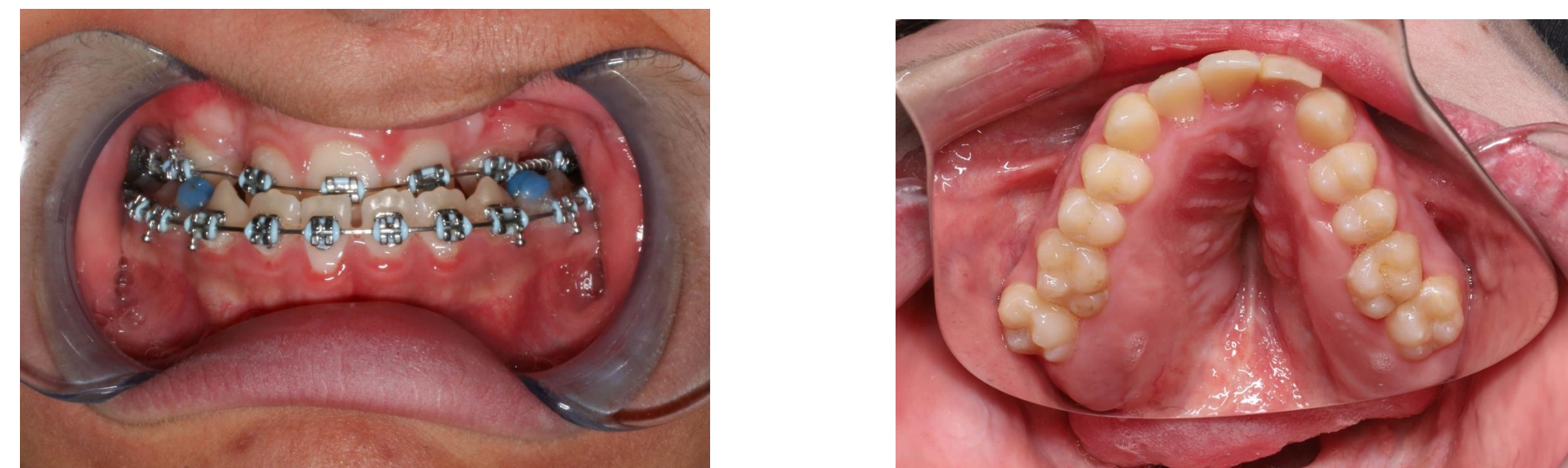
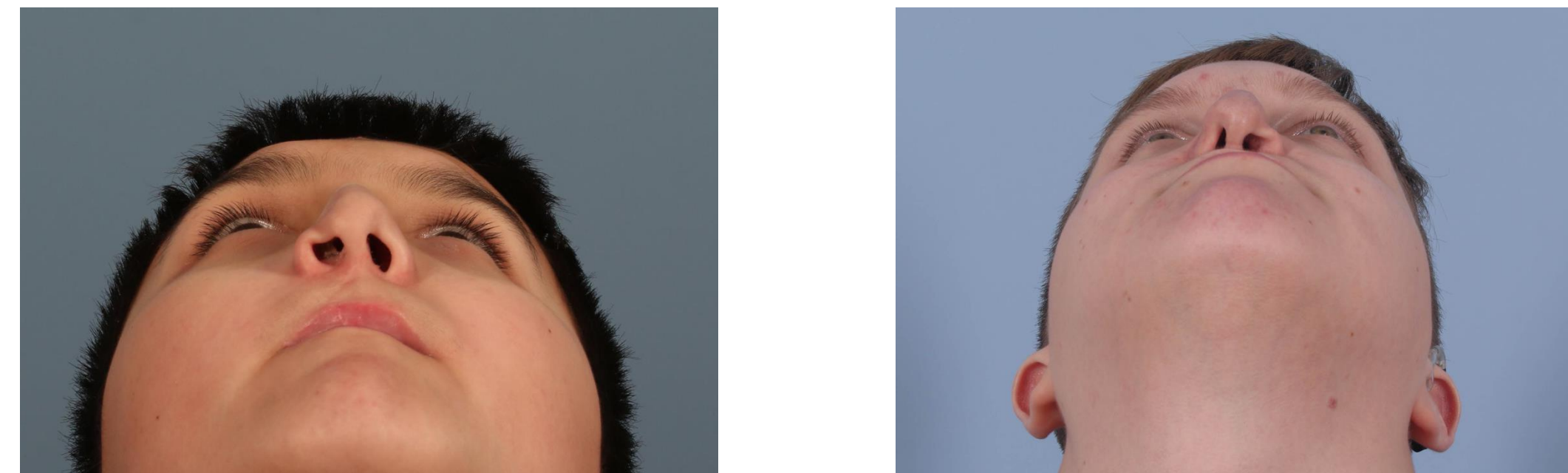


## ORAL HEALTH IMPLICATIONS

Individuals with CHARGE syndrome frequently present with a range of congenital dental anomalies, including taurodontism, hypodontia, ectopic eruption, submergence of primary molars, and variations such as agenesis or supernumerary teeth. Less common but clinically significant findings include cleft lip or palate, micrognathia, ogival palate, and facial palsy, all of which may influence oral function and treatment planning. Cleft lip is more prevalent than cleft palate in this population, and when clefting is present, the choanae are typically patent; in some cases, clefting may substitute for choanal atresia in diagnostic criteria. Submucous cleft palate is often diagnosed later in childhood, particularly following tonsil or adenoid removal. Dental management requires careful consideration of systemic comorbidities: cardiac defects present in two-thirds of patients may affect procedural safety, while airway anomalies such as laryngomalacia or tracheomalacia complicate sedation and anesthesia. Cranial nerve palsies can impair feeding and increase aspiration risk postoperatively. Additionally, hearing and vision impairments necessitate modified communication strategies, desensitization techniques, and close collaboration with caregivers to ensure safe and effective dental care.

## DIAGNOSIS AND SYSTEMIC FINDINGS

Diagnosis of CHARGE syndrome is guided by the classic **3C triad**—coloboma, choanal atresia, and abnormal semicircular canals—with additional supportive features such as orofacial clefting, distinctive facial appearance, tracheoesophageal fistula, and limb anomalies. Genetic confirmation is typically achieved through identification of a *CHD7* mutation on chromosome 8q12. Systemic involvement is extensive and variable: ocular findings range from iris coloboma to retinal involvement that may impair vision or predispose to retinal detachment, while cranial nerve dysfunction occurs in 70–90% of cases and may manifest as optic nerve coloboma, unilateral facial palsy, sensorineural hearing loss, and significant swallowing or oromotor difficulties. Cardiac anomalies—including tetralogy of Fallot, ventricular septal defects, atrioventricular canal defects, and aortic arch abnormalities—are common and clinically significant. More than half of affected children exhibit choanal atresia, contributing to respiratory compromise. Growth restriction typically emerges in infancy, with many children falling below the 3rd percentile due to feeding challenges and medical complexity, and delayed puberty is seen in over 70% of cases. Genital abnormalities such as micropenis, undescended testes, or hypoplastic labia are frequently present. Ear anomalies are distinctive and diagnostically helpful, characterized by triangular conchae, short and wide cupped ears, and discontinuity of the antihelix and antitragus, while semicircular canal hypoplasia with vestibular areflexia is considered a constant feature.



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