Depressed Nasal Bridge in Pediatric Orthopaedic Practice: A Review

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Abstract
Depressions of the nasal bridge may be localized on the bone, cartilage or both. The latter are the most severe cases constituting the saddle-nose deformity, which may lead to various serious functional consequences. A severely depressed nasal dorsum is usually secondary to trauma or infection. In children it may also be caused by a multitude of genetic or environmental factors. A wide variety of congenital disorders or syndromes, birth defects, such as the fetal alcohol syndrome, and infectious diseases, such as congenital syphilis may be included in the pathogenetic factors. The most commonly encountered congenital syndromes with a depressed nasal bridge in the pediatric orthopaedic practice are cleidocranial dysplasia, children with neurodevelopmental delay, achondroplasia, Conradi-Hünermann-Happle syndrome, Cornelia de Lange syndrome, osteogenesis imperfecta and Klippel-Feil syndrome.

Review

The depth of the nasal bridge is evaluated from the profile view. The severity of nasal bridge depression as well as the associated clinical findings may vary considerably. In the mild forms the defect causes mainly aesthetic problems, while in the most advanced cases severe airway obstruction and inability to feed may occur. The characteristics of a child are less developed at birth, and with development the nose bridge is likely to acquire a more normal appearance. Saddle depression of the nasal bridge is one of the most common nasal deformities showing varying degrees of severity. It describes nasal profile resembling a riding saddle. It may be relative or true. Relative is when there is a hump formation or excessive projection of the nasal tip or both. In true saddle deformity, there is actual loss of tissues along the dorsal nasal line. Saddle-nose deformity may occur as a result of trauma to the nose (craniofacial trauma, injuries-the boxer’s nose) or as a complication of nasal surgery. It may also be caused by specific infections, such as syphilis, tuberculosis, leprosy, leishmaniasis and other non-specific suppurative infections. In some cases, saddle nose develops from chronic nasal inflammation caused by disorders such as relapsing polychondritis and granulomatosis with polyangiitis (Wegener’s granulomatosis). Habitual use of cocaine or inhaling other drugs may cause saddle-nose deformity. Since these deformities may also arise without an evident precipitating cause, they can pose a diagnostic dilemma. The progressing saddle-nose deformity may also be due to sarcoidosis and tumor invasion.

The congenital low nasal bridge or the saddle-nose deformity is a rare occurrence in the pediatric orthopaedic practice. The definition indicates that the deformity is present at birth. The causes may include a birth defect, such as the fetal alcohol syndrome, an infectious disease, such as congenital syphilis, and inherited disorders, such as the ectodermal dysplasias, congenital anomalies due to nose un-
derdevelopment and congenital syndromes, such as Dysostosis cleidocranial, Williams syndrome, Down syndrome, Achondroplasia, Conradi-Hünermann-Happle syndrome, Cornelia de Lange syndrome, Osteogenesis imperfect and Klippel-Feil syndrome. Studies to detect genetic abnormalities or other health problems may include x-rays to estimate the structure of the child’s nose, chromosome tests to detect genetic abnormalities and blood tests to check the level of enzymes and inflammatory markers [1-17].

**Birth defects**

Fetal alcohol syndrome indicates a wide variety of birth defects associated with the use of alcohol during the first trimester of pregnancy. It may be complicated by nervous system or behavioral problems, facial abnormalities, such as a depressed nasal bridge, growth deficiencies and learning disabilities. Alcohol is the most frequent and most important teratogenic noxa for the embryo and fetus [18-21].

**Infectious diseases**

Congenital syphilis is a severe and potentially life-threatening infection in infants caused by *Treponema pallidum* transmitted by infected mother to her baby almost exclusively via the placental barrier after the fourth month of pregnancy. Perinatal infections are less frequent, and postnatal infections are only exceptionally encountered. The symptoms of congenital syphilis may be divided into prenatal (syphilis materno-fetalis), neonatal, and rarely seen postnatal. Babies born with congenital syphilis frequently have no bridge to nose and severe congenital pneumonia. Associated health problems may include blindness, deafness, neurological problems and skeletal manifestations, including periostitis, osteitis, metaphyseal changes (Figure 1), pseudoparalysis, pathological fractures, joint involvement and dactylitis. Penicillin is the only antibiotic of proven value for the treatment of congenital syphilis [22-26].

**Inherited disorders**

Because of the saddle-nose deformity and bilateral cataracts all patients suspected of having congenital syphilis should be investigated for ocular or auditory defects, which would confirm the diagnosis of ectodermal dysplasia. Ectodermal dysplasias are a large and complex group of inherited disorders characterized by deficient ectodermal and mesodermal development. Nasal obstruction due to the presence of nasal crusting, hearing loss and throat hoarseness are the most represented symptoms [27-30].

Congenital anomalies of the nose include a broad spectrum of defects, being a result of abnormalities in the developmental process. These conditions range from partial deformities of the nose, such as isolated absence of the nasal bones, absence of the columella, absence of the septal cartilage or alae cartilage, through hemi aplasia of the nose to complete absence of the nose (arhinia) [17].

**Congenital syndromes**

The nasal bridge may be depressed, or lie deeper in the face than normal, in various craniosynostosis syndromes and skeletal dysplasias. The most common congenital skeletal dysplasias that may be associated with a low nasal bridge in the pediatric orthopaedic practice are:

1. Cleidocranial dysplasia is a well defined skeletal and dental disorder with characteristic clinical findings and autosomal dominant inheritance. Major indicators of the disease include hypoplasia or aplasia of clavicular bones. Bilaterality is the rule but not always the case (Figure 2). The missing segment may be represented by fibrous pseudarthrosis or by a fibrous tether or cord. Craniofacial growth is affected in many ways and may include a depressed nasal bridge. The thoracic cage is small and bell shaped with short, oblique ribs. The pelvis is invariably involved and shows characteristic changes. A relatively constant abnormality
is the presence of both proximal and distal epiphyses in the second metacarpals and metatarsals leading to excessive growth and length. All other bones of the hands and feet, especially the distal phalanges and the middle phalanges of the second and fifth fingers are unusually short. Final height is significantly reduced. Congenital pseudarthrosis of the clavicle is probably among the most common conditions to be differentiated. It was initially described in association with cleidocranial dysplasia. In the great majority of cases involvement is unilateral with a marked predominance of the right side. The cases are sporadic and there is no other bone involvement [31-35].

2. Neurodevelopmental delay and dysmorphic facial features require a thorough evaluation and extensive diagnostic testing in children (Figure 3). A) Williams syndrome is a developmental disorder caused by a hemizygous contiguous gene deletion on chromosome 7q11.23. It is characterized by distinct facial features, congenital heart disease, mental retardation and a gregarious personality. It may also be complicated by bone deformities like a depressed nose [36-39]. B) Down syndrome also known as trisomy 21, is the most common chromosomal abnormality among live born infants reaching up to 1 in 700 births. It is typically associated with physical growth delays, characteristic facial features and mild to moderate intellectual disability. It is characterized by a variety of dysmorphic features and medical conditions. The former include a small chin, slanted eyes, poor muscle tone, a flat nasal bridge, a single crease of the palm, and a protruding tongue due to a small mouth and relatively large tongue. The latter include poor immune function, congenital heart defect, atlantoaxial instability, epilepsy, leukemia, thyroid diseases, and mental disorders. Children with Down syndrome are at increased risk of ear-nose-throat disorders such as refractory otitis, eustachian tube dysfunction, laryngomalacia, tracheal stenosis, obstructive sleep apnea, hearing loss, as well as voice and articulatory impairments [40-44].

3. Achondroplasia is a human bone genetic disorder of the growth plate and is the most common form of inherited disproportionate short stature. It is inherited as an autosomal dominant disease with essentially complete penetrance. Of these most have the same point mutations in the gene for fibroblast growth factor receptor 3, which is a negative regulator of bone growth. The clinical and radiological features of achondroplasia may easily be identified; they include disproportionate short stature with rhizomelic shortening, lumbar hyperlordosis, and a trident hand configuration. Achondroplasia is also of dental interest because of its characteristic craniofacial features which include relative macrocephaly with frontal bossing (prominent or bulging forehead with a depressed nasal bridge), midface hypoplasia and maxillary hypoplasia (Figure 4a and Figure 4b). The majority of achondroplasts have a normal intelligence, but many social and medical complications may compromise a full and productive life. Some of them have serious health consequences related to hydrocephalus, craniocervical junction compression, or upper-airway obstruction [45-49].
4. The Conradi-Hünermann-Happle syndrome is characterized by dysmorphic face with depressed nasal bridge, skin manifestations, heart defects, cataract, hypotonia, short stature, kyphoscoliosis, coxa vara, upper cervical instability and stippled calcifications within the epiphyses. Chondrodysplasia Punctata (CDP) is a rare, heterogeneous congenital skeletal dysplasia, characterized by punctate or dot-like calcium deposits in cartilage observed on neonatal radiograms (Figure 5). A number of inborn metabolic diseases are associated with CDP, including peroxisomal and cholesterol biosynthesis dysfunction and other inborn errors of metabolism such as: mucolipidosis type II, mucopolysaccharidosis type III, GM1 gangliosidosis. CDP can be seen in several disorders and syndromes such as Turner, Zellweger, trisomy 21, trisomy 18, FAS and following ingestion of warfarin or phenytoin during pregnancy. The inheritance transmission may be autosomal dominant type, autosomal recessive type, X-linked dominant type, X-linked recessive type with deletion of terminal short arm or duplication of short arm of chromosome 16 [50-67].

5. Cornelia de Lange syndrome (also called Bushy syndrome or Amsterdam dwarfism), is a genetic disorder that can lead to several alterations. This disease affects both physical and neuropsychiatric development. The various abnormalities include facial dysmorphia (arched eyebrows, synophrys, depressed nasal bridge, long philtrum, down-turned angles of the mouth), upper-extremity malformations, hirsutism, cardiac defects, and gastrointestinal alterations. Classical de Lange syndrome presents with a striking face, pronounced growth and mental retardation, and variable limb deficiencies (Figure 6a and Figure 6b). Over the past five years, a mild variant has been defined, with less significant psychomotor retardation, less marked pre- and postnatal growth deficiency, and an uncommon association with major malformations, although mild limb anomalies may be present [68-76].

6. Osteogenesis Imperfecta (OI) or brittle bone disease is a rare hereditary disorder of type 1 collagen synthesis with a triad of clinical features including multiple fractures, blue sclera and conductive hearing loss. It exhibits wide variation in appearance and severity. The most severe forms lead to early death. Clinical features such as fracture frequency, long bone deformities (Figure 7a), muscle strength or extraskeletal problems vary widely. Some features are age dependent. The initial classification by Looser described two types: OI congenita and OI tarda. Sillence and
colleagues described 4 types of the disease taking into account the phenotypic features and the mode of inheritance. Scleral hue is an important sign which distinguishes 2 broad groupings of patients, those with and those without blue sclera, with nonlethal OI. Individuals with OI type I (Figure 7b) have distinctly blue sclerae which remain intensely blue throughout life. In OI type III and OI type IV the sclerae may also be blue at birth and during infancy, but the intensity fades with time such that these individuals have sclerae of normal hue by adolescence and adult life. In children with the most severe nonlethal forms of osteogenesis imperfecta severe craniofacial and dental anomalies may be encountered, although there are only very few reported OI patients treated with rhinoplasty [77-81].

7. The Klippel-Feil syndrome may be associated with osseous and visceral anomalies. It usually presents with a clinical triad including a short tilted neck, low posterior hairline, and loss of the cervical motion. It is characterized by the fusion of at least two cervical vertebrae. An unrecognized fetal alcohol syndrome has been questioned in the pathogenesis, although the two syndromes are distinct entities. A variety of craniofacial anomalies, including a depressed nasal bridge (Figure 8), may occur, in association with the syndrome, widening its clinical spectrum [82-92].
Depressed nasal bridge is a rare and probably underestimated condition in neonates. It may be due to a wide variety of disorders with completely different phenotype and genotype. The pathogenic pathway in causing the specific anomaly is completely different in each disorder. Pediatric consultation through an early referral is essential for a successful early identification, prompt diagnosis and therapy of infants with a depressed nasal bridge or a saddle-nose deformity due to birth defects, infectious diseases, inherited disorders and congenital syndromes.

Conflict of Interest Statement

The author certifies that he has no commercial associations (such as consultancies, stock ownership, equity interest, patent/licensing arrangements, etc) that might pose a conflict of interest in connection with the submitted article. The author received no financial support for this study.

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