

Frontonasal dysplasia: A case report

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Frontonasal dysplasia is an uncommon congenital anomaly with diverse clinical phenotypes and highly variable clinical characteristics, including hypertelorism, a broad nasal root, median facial cleft, a missing or underdeveloped nasal tip, and a widow's peak hairline. Frontonasal dysplasia is mostly inherited and caused by the *ALX* genes (*ALX1*, *ALX3*, and *ALX4*). We report a rare case of a frontonasal dysplasia patient with mild hypertelorism, a broad nasal root, an underdeveloped nasal tip, an accessory nasal tag, and a widow's peak. We used soft tissue re-draping to achieve aesthetic improvements.

Keywords: Frontonasal dysplasia / Frontorhiny / Median facial cleft

INTRODUCTION

Frontonasal dysplasia is a rare genetic disorder resulting in physical malformations encompassing a broad range of face and head anomalies. Its major clinical characteristics include ocular hypertelorism, a broad nasal root, median facial cleft, an underdeveloped or absent nasal tip, and a widow's peak hair line [1]. Features such as ocular changes, intellectual disability, ankyloglossia, accessory nasal tag, cleft lip, and deafness have occasionally been reported [2,3].

Although the mode of inheritance remains unclear, frontonasal dysplasia has been suggested to have an autosomal recessive or autosomal dominant pattern of inheritance. It has mostly been reported in sporadic cases, and can manifest in syndromic or non-syndromic forms [4]. The etiology of dysplasia remains unknown, but it is thought to result from abnormal development of the frontonasal prominence in craniofacial embryogenesis [5].

Frontonasal dysplasia is a heterogeneous anomaly that is associated with a diverse range of genetic causes. Only a small number of cases have been reported and treated. We present the

case of a patient with frontonasal dysplasia characterized by mild hypertelorism, a broad nasal root, an underdeveloped nasal tip, an accessory nasal tag, and a widow's peak. We treated the nasal deformity by soft tissue re-draping and observed aesthetic improvements.

CASE REPORT

A 9-year-old boy presented with mild hypertelorism and a nasal anomaly. He had a broad nasal root, an underdeveloped nasal tip, a short columella, an accessory nasal tag, and a widow's peak (Fig. 1). He was of normal intelligence, and a clinical examination did not detect ocular anomalies, hearing loss, cleft lip, or ankyloglossia. His parents and siblings were not affected and presented a normal phenotype. There was no familial history of frontonasal dysplasia.

Due to economic reasons, a further evaluation was not possible. The patient's parents only wanted to improve his appearance. We decided to manage the accessory nasal tag and deformed cartilage for aesthetic improvement. We made an incision between the concave lesion and the convex lesion of the skin tag. Upon exploration, the entire nasal cartilage appeared normal structurally, but all areas of cartilage were thin, weak, and flat. We cinched the medial alar cartilage and performed intradermal suturing to strengthen the strut. The flaring lateral

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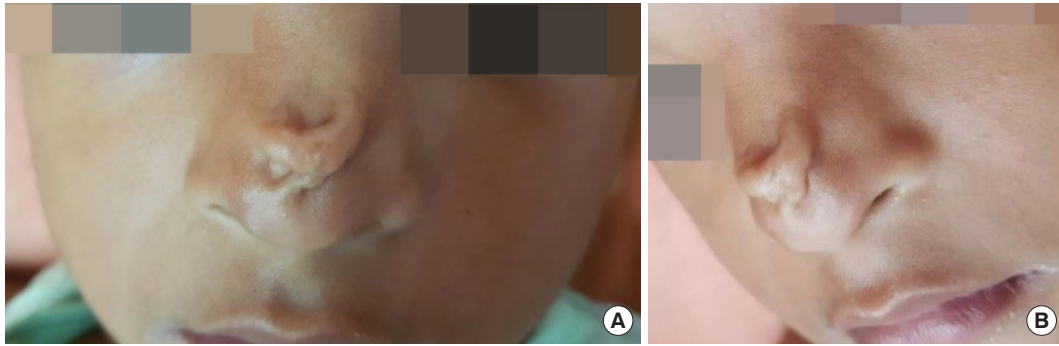


Fig. 1. (A, B) Preoperative photograph of a 9-year-old boy. Hypertelorism, an accessory nasal skin tag, an underdeveloped nasal tip, a short columella, and a widow's peak were present.

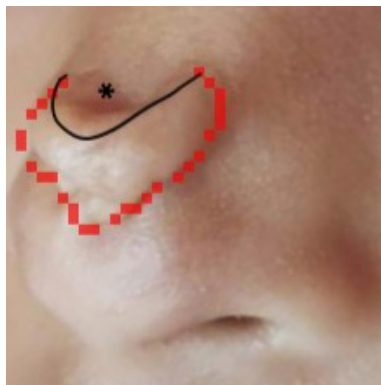


Fig. 2. Surgical plan. Black line, incision line; red dotted line, excision margin of remnant skin; asterisk, skin area where the incision would be released.

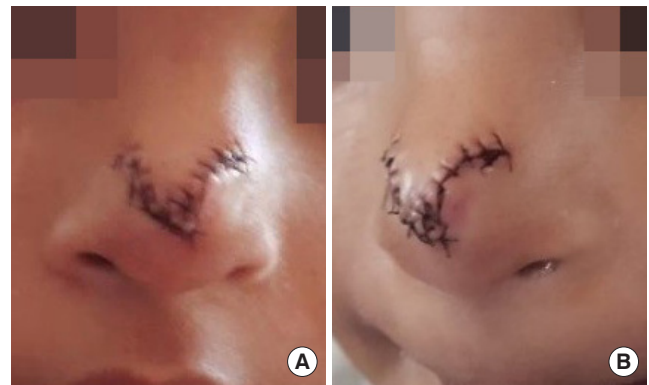


Fig. 4. (A, B) Postoperative photographs showing an improved appearance.

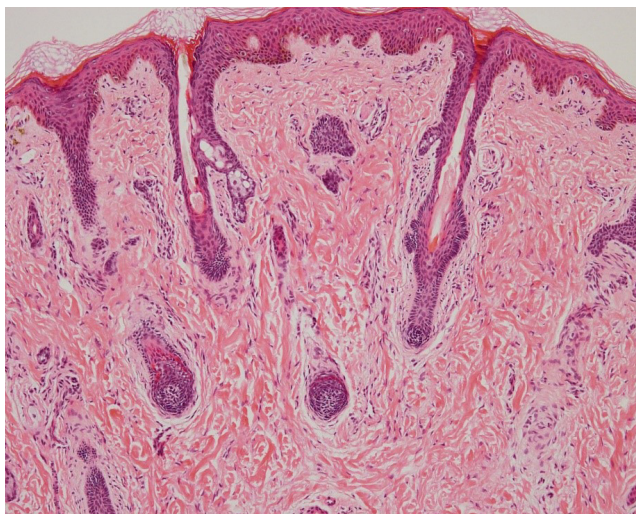


Fig. 3. Histopathology of the skin tag, revealing an increased amount of dense irregular connective tissue in the dermis (H&E, ×100).

alar cartilage was corrected using sutures. We decided to use the skin of the concave lesion to cover the defect site where the accessory skin tag was located. We straightened the skin by releasing the incision on the dermis, and re-draped it over the de-

fect site (Fig. 2).

The skin tag had a soft rubber-like texture, and its histopathologic findings revealed enriched collagen and a fibrous tissue layer (Fig. 3). The postoperative photographs showed aesthetic improvements in the patient's appearance (Fig. 4). However, the patient still had a very short columella and unusually wide alar cartilage. We recommended a follow-up operation at around 14 years of age, after the patient will have reached skeletal maturity.

DISCUSSION

Frontonasal dysplasia is an unusual genetic malformation that presents with a variety of clinical phenotypes; nonetheless, its major characteristic manifestations include ocular hypertelorism, a broad nasal root, a median facial cleft, an underdeveloped or an absent nasal tip, and a widow's peak hairline [1]. Frontonasal dysplasia is diagnosed on the basis of presentation with at least two major clinical features. Additional manifestations of frontonasal dysplasia include ocular changes, intellectual disability, ankyloglossia, an accessory nasal tag, cleft lip, hearing loss, and agenesis of the corpus callosum [2,3].

The etiology of frontonasal dysplasia has yet to be established conclusively, but it is thought to be a type of developmental field defect. Specifically, frontonasal dysplasia is believed to be an abnormal development of the frontonasal prominence in craniofacial embryogenesis [5]. Frontonasal dysplasia was first described by Sedano and Gorlin [5], who emphasized that it involves a primary medial facial defect. In the embryonic development of the face, the persistence of the frontonasal process in its position prevents the orbits from reaching their normal position. This is thought to cause the distinct hypertelorism characteristic of this condition, as well as nasal deformities and cleft lip.

Twigg et al. [6] reported three cases with the characteristic clinical features of frontonasal dysplasia and identified the causative gene of frontonasal malformation as *ALX3*, a homeobox gene. They used the term “frontorhiny” to refer to this malformation, and found it to be an inherited, autosomal recessive condition. Lees et al. [7] reported 11 additional patients from seven families. A genetic analysis showed mutations involving the same *ALX3* homeobox gene.

Frontonasal dysplasia is mostly sporadic, and it has been reported to manifest as syndromic (i.e., in association with other severe anomalies) or as non-syndromic. Three types of frontonasal dysplasia have been identified, depending on the specific *ALX* gene in which a mutation occurs (*ALX1*, *ALX3*, or *ALX4*). Frontonasal dysplasia caused by *ALX1* or *ALX4* is inherited in an autosomal recessive or autosomal dominant fashion, respectively, and has a more severe manifestation than other forms of frontonasal dysplasia [8].

As only a few cases of frontonasal dysplasia have been reported, its treatment strategy is not well-established. Multistage craniofacial surgery is needed. If craniosynostosis is present, primary craniosynostotic correction is performed at the age of 3 to 6 months. Subsequently, orbital hypertelorism is corrected at the age of 5 to 6 years, after maxillary central incisor eruption. Nasal anomaly correction is either performed simultaneously with the correction of hypertelorism or after the patient achieves skeletal maturity, at the age of 14 to 17 years [9].

If the deformity persists, revision surgery or secondary rhinoplasty is required. Surgery may be suggested for patients older than 14 years of age [9]. Due to the psychological trauma experienced when a patient becomes aware of the deformity and the subsequent impact on self-esteem, no absolute consensus exists on the timing of the operation. As patients become aware of deformities at 3.5 years or later, corrective operations are recommended at as early as 2 years of age [10].

Frontonasal dysplasia is a rare sporadic congenital deformity. For this reason, few cases have been reported. Herein, we describe the case of a patient with frontonasal dysplasia who pre-

sented with typical features, including hypertelorism, a broad nasal root, an underdeveloped nasal tip, a short columella, an accessory nasal tag, and a widow’s peak, and review the patient’s treatment. The operation improved the patient’s physical appearance.

NOTES

Conflict of interest

No potential conflict of interest relevant to this article was reported.

Ethical approval

The study was performed in accordance with the principles of the Declaration of Helsinki. Written informed consent was obtained.

Patient consent

The patient’s parent provided written informed consent for the publication and the use of his images.

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