CASE REPORT

Ortho-surgical management of Arhinia syndrome: A case report

Prise en charge ortho-chirurgicale du syndrome d'Arhinia: A propos d'un cas

Abdelali Halimi[1], Fatima Zaoui[1], Mohamed El-Okeily[2], Nawal Bouyahyaoui[1]

1. Mohammed V University in Rabat, Faculty of Dentistry, Department of Orthodontics and dentofacial orthopedics, Rabat, Morocco.
2. Aesthetic surgeon, private liberal activity, SOFCPRE - French Society of Plastic, Reconstructive and Aesthetic Surgery, Bordeaux, France.

Abstract

Introduction and importance: Arhinia syndrome (AS) is an extremely rare malformation characterized by typical clinical signs such as a lack of nose, with difficulty in breathing and inability to feed due to airway obstruction. The ortho-surgical management of AS is a big challenge and it requires an interdisciplinary relationship between the orthodontist and a maxillofacial surgeon. The authors reported a case of AS.
Case presentation: The only case in Morocco among fewer than 30 cases described in the whole world, male child, 7-year-old.
Clinical findings and investigations: Complete absence of the external nose, nasal cavities, and other characteristic signs. This diagnosis was genetically confirmed.
Interventions and outcome: A LeFort II maxillary osteotomy was done in combination with a facemask and miniscrew followed by a parietal bone graft and maxillary expansion with a customer expander. An internal and external nasal reconstruction was undergone with orthodontic management of unerupted teeth. An acceptable morphology for the newly created external nose was obtained, this reconstruction was viable and esthetically acceptable. A slight external restenosis of a left nave was observed. This result was stable over three years.
Relevance and impact: AS is a rare condition with a pathogenesis not been fully understood. The ideal treatment should have the lowest possible morbidity and a low relapse rate. The most difficulty in the approach is the creation and maintenance of a nasal cavity. These therapeutic aspects will be discussed through this case. The use of facemask therapy with a miniscrew potentiated the effect of osteotomy and stabilized the parietal bone graft which allowed nasal reconstruction and orthodontic management of unerupted teeth in this case.

Keywords: Case report, Ortho-surgical management, Arhinia syndrome

Résumé

Introduction et importance: Le syndrome d'Arhinia (SA) est une malformation extrêmement rare caractérisée par des signes cliniques typiques tels que l'absence de nez, des difficultés respiratoires et une incapacité à s'alimenter due à une obstruction des voies aériennes. La prise en charge ortho-chirurgicale de cette anomalie représente un défi majeur et nécessite une collaboration interdisciplinaire entre l'orthodontiste et un chirurgien maxillo-facial, ce propos est illustré ici par ces cas d'Arhinia.
Présentation du cas: Le seule cas au Maroc parmi moins de 30 cas à travers le monde entier, un garçon de 7 ans.
Constatations cliniques et investigations: Absence complète de nez externe, de fosses nasales et d'autres signes caractéristiques. Ce diagnostic a été confirmé génétiquement.
Interventions et résultats: Une ostéotomie maxillaire de LeFort II a été réalisée en association avec un masque facial supporté par une gouttière maxillaire tenue par une mini-vis, suivie d'une greffe osseuse pariétale et d'une expansion maxillaire chirurgicale. Une reconstruction nasale interne et externe a été réalisée avec prise en charge orthodontique des dents incluses. Une morphologie externe acceptable du nez a été obtenue, cette reconstruction nasale était viable et esthétique. Ce résultat est resté stable sur trois ans. Une légère sténose externe du nez gauche a été observée.
Pertinence et impact: Le SA est une affection rare dont la pathogénèse reste mal comprise. Le traitement idéal devrait présenter une morbidité minimale et un faible taux de récidive. La principale difficulté réside dans la création et le maintien d'une fosse nasale. Ces aspects thérapeutiques seront abordés à travers ce cas. L'utilisation d'un masque facial avec gouttière et mini-vis a potentialisé l'effet de l'ostéotomie et stabilisé la greffe osseuse pariétale, ce qui a permis la reconstruction nasale et la prise en charge orthodontique des dents incluses dans ce cas.

Mots-clés: Prise en charge orthodontico-chirurgicale, Rapport de cas, Syndrome d'Arhinia

Correspondance
Abdelali Halimi
Mohammed V University in Rabat, Faculty of Dentistry, Department of Orthodontics and dentofacial orthopedics, Rabat, Morocco.
Email: a.halimi@um5r.ac.ma

LA TUNISIE MEDICALE-2025; Vol 103 (08): 1122-1127 DOI: 10.62438/tunismed.v103i8.5947

Introduction

Arhinia syndrome (AS) is an extremely rare malformation and there have been extremely few cases reported, which characterized by typical clinical signs such as a lack of nose, with difficulty in breathing and inability to feed due to airway obstruction (1). The lack of an external nose is generally one part of a complex malformative syndrome characterized by several clinical signs (2). The etiology of arhinia is unknown and the pathogenesis of this disease has not been fully understood (3). It is postulated that lack of development of the nose results from medial failure and lateral nasal process growth, but it is also possible that overgrowth and premature fusion of the nasal medial process result in the formation of the atretic plate (4). Genetic correlations are inconsistent; and most cases are sporadic. The literature includes familial cases (5). No single gene or chromosome has been identified as being responsible for this malformation (6). Gordon reported missense mutations in the epigenetic regulator SMCHD1 (encoding structural maintenance of chromosomes flexible hinge domain containing 1; NM_015295.2) mapping to the extended ATPase domain of the encoded protein cause Bosma arhinia microphthalmia syndrome (BAMS) in 14 cases studied (6). All mutations were de novo where parental DNA was available and SMCHD1 has a key player in nasal development and provide biochemical insight into its enzymatic function that may be exploited for development of therapeutics for facioscapulohumeral muscular dystrophy (FSHD) (7).

The pathophysiology of arhinia is poorly understood, and various theories have been proposed (4). Reduced growth of the medial and lateral nasal processes, excess growth of the medial nasal processes, defective reabsorption of the epithelial plates, and arrested migration of neural crest cells are all possible explanations for these malformations (7). Because AS is exceptionally rare and few cases have been treated, no standardized treatment protocol is available (4).

The main objective of our paper was to present the course of a combined ortho-surgical approach of a child with AS because very few of the published cases were treated with a global approach and this is the only case in Morocco among fewer than 30 cases described in literature worldwide (7). This report has been written in accordance with the Surgical Case Report Criteria guidelines for case reports (8).

Patient information

A male child, 7-year-old was referred by a colleague for orofacial rehabilitation. The child was born in Morocco to healthy unrelated parents. There was neither a family background of congenital malformations nor a history of any medications during the pregnancy. The prenatal time was uncomplicated. The child showed a syndromic presentation compatible with AS or BAMS.

Primary concerns and symptoms of the patient: the child showed complete absence of the external nose, nasal cavities. This child had difficulty in breathing and inability to feed due to airway obstruction. The child showed a Class III malocclusions.

Genetic analysis for the case led to the identification of de novo heterozygous missense mutations in the SMCHD1 gene, which were confirmed by Sanger sequencing. Heterozygous missense mutation was identified in this case and the variants were de novo, suggesting germline mutations in parental gametes (6).

Clinical Findings

A normal clinical examination revealed a complete absence of the external nose, nasal cavities, paranasal sinuses, and olfactory apparatus and defects causing a difficulty in breathing and inability to feed due to airway obstruction. We observed the presence of a very thinner protuberances in the midface, associated, hypertelorism, microphthalmia, colobomas, nasolacrimal duct abnormalities, maxillary anteroposterior, vertical, and transverse hypoplasia (midface hypoplasia), high arched palate. Clinical profile examination revealed a transfrontal profile, maxillary hypoplasia, a slightly increased cervical-chin distance, a marked labiochin line, and absence of nose and nasolabial angle. An oral examination revealed moderate oral health with no periodontal problems, severe crowding, enamel demineralization and delayed tooth eruption in mixed dentition. The interarch relationship showed a Class III malocclusions, serious bilateral crossbite with open bite. A temporomandibular joint examination revealed no centric occlusion discrepancy with no history of any pain or discomfort in the temporomandibular joint or associated muscles (Figure 1 (a and b)).

Timeline of current episode

The patient initially presented with a 7-year history of a complete absence of the external nose, and olfactory apparatus and defects causing a difficulty in breathing and inability to feed due to airway obstruction. The confrontation of clinical and radiological signs confirmed the diagnosis of AS. At age ten years, he was treated with Le Fort II maxillary osteotomy with a bicortical approach under submental intubation. The facemask was applied for six months. This mask was supported by a resin splint screwed into the hard palate. After six months, a parietal bone graft to fill the distracted zone. The patient continues to wear the facemask. At age 11, a nasal reconstruction was done and two nasal tubes were placed in the newly created nares which allow breathing and held in place for six months. The transverse expansion was started by a tri-helix manually shaped. Post-operatively, the nasal tube was changed from 3mm size to 5.5mm size under local care, and the tri-helix has been replaced by a Hyrax expander. At the six month follow-up, the patient reported had no difficulty breathing and eating due to clear airways. At the latest follow-up, the patient reported continued satisfaction with the outcome and had resumed his activities normally (Table 1).

A horizontal arm linked the two sides. Then the maxilla was down fractured. The effect of this osteotomy was

Table 1. Timeline of events and interventions.

Patient age (years, y, month, m)	Event/Intervention	Details
6 y	Genetic analysis	Identification of *de novo* heterozygous missense mutations in the SMCHD1 gene
7 y	Diagnosis of Arhinia syndrome (AS)	The child was initially referred by a colleague for orofacial rehabilitation. Clinical and radiographic examination confirmed the diagnosis of AS.
10 y	Surgical intervention 1st step	A Le Fort II maxillary osteotomy was done with a bicortical approach under submental intubation. The facemask was applied for 6 months. This mask was supported by a resin splint screwed into the hard palate.
10 y 6 m	Surgical intervention second step	A parietal bone graft was done to fill the distracted zone. The patient continues to wear the facemask.
11 y	Surgical intervention 3rd step	A nasal reconstruction was done and two nasal tubes were placed in the newly created nares which allow breathing and held in place for 6 months. The transverse expansion was started by a tri-helix manually shaped.
11 y 6 m	Post-operative care	The nasal tube was changed from 3mm size to 5.5mm size under local care and the tri-helix has been replaced by a Hyrax expander.
12 y	Follow-up	The patient reported had no difficulty breathing and eating due to clear airways.
13 y	Follow-up	The patient reported continued satisfaction with the outcome and had resumed his activities normally.
13 y 6 m	Follow-up	Currently we are continuing our orthodontic treatment.

Diagnostic assessment

A panoramic radiograph showed a missing maxillary sinus, and ethmoid bone, and the proper bones of the nose, deep unerupted teeth and a sever crowding in a mixed dentition, no bone pathology, and no temporomandibular pathology were observed. Cephalometric examination showed a skeletal Class III malocclusions with skeletal relationship and maxillary retrognathism, Angle Class III malocclusion, bilateral crossbite, serious open-bite, asymmetrical occlusal relationship, severe crowding, enamel demineralization and delayed tooth eruption in mixed dentition (Figures 1 (b, c, d)).

Computed tomography (CT) shows a facial malformation with partial agenesis of the maxilla and ethmoid and confirmed the absence of the internal nasal cavity and paranasal sinuses, absence of nasal pyramid with integrity of the orbital frames and respect for the mandible (Figure 1e). The prenatal time was uncomplicated.

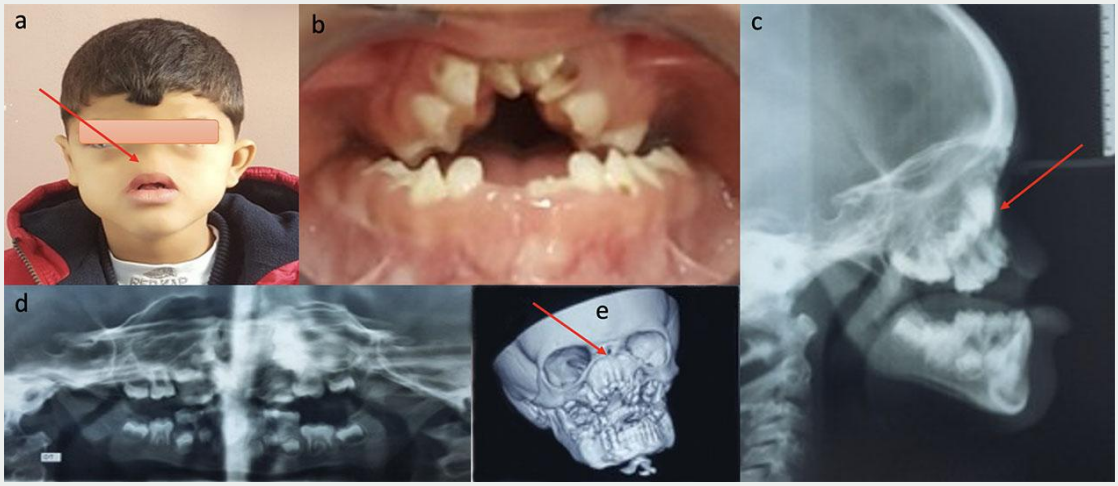

Figure 1. Pretreatment extraoral **(a)**, and intraoral photographs **(b)**, and radiographs **(d, c, e)**. The arrow in figure **(a)** shows a complete absence of the external nose and in figure **(c and e)** shows the absence of the internal nasal cavity and paranasal sinuses.

Diagnosis

The final diagnosis was AS or BAMS.

Therapeutic interventions

The therapeutic objectives of this case were as follows: (1) Preparation of the grafting site for the nasal cavity and creation of an internal and external nose with lowest morbidity; (2) maintenance of a nasal cavity with functional and esthetic aims and a low relapse rate; (3) ortho-surgical management of impacted teeth; and (4) orthodontic management of class III malocclusion, crossbite and crowding.

The patient underwent a Le Fort II maxillary osteotomy with a bicortical approach under submental intubation. This osteotomy was done to give enough height of maxillary. This was extended through the maxilla-zygomatic consoles and toward the medial end of the inferior orbital edge and ran through the infraorbital foramen.

potentiated by the facemask performed in the operating room. The facemask was applied force at a 25-30° angle to the occlusal plane in a down ward direction for six months. The facemask allowed external distraction and provides additional facial height in a reasonable time period and has sufficient midfacial vertical length to accommodate a nasal reconstruction and suitable aesthetic proportions. This mask was supported by a resin splint screwed into the hard palate. These splint carries two shaped ratchets on each side which support extra-oral elastic (Figure 2 (a, b)). The magnitude of the force was 500g per side. The duration of the force was 24h/day for 12 weeks (9).

In a second step, the young patient benefited from a parietal bone graft to fill the distracted zone. The patient continues to wear the facemask. In a third step, the patient underwent a nasal reconstruction. A wide median nasal cavity was created with a round bur, reaching posteriorly to the upper portion of the rhinopharynx and then extend it along the maxillary bone. After the creation of the new nasal cavity and to avoid the closure of this cavity especially by blood in the beginning and bone secondly, it was necessary to let a nasal tube which allow breathing. Naturally the mucosa cells coming from oro-pharyngeal area, colonize the bone surface of this new nasal cavity. Then, we change the nasal tube increasing its diameter. For this case we started with the 3mm size tube then moved to the 5.5mm size. Even we know that it will be really tough to obtain a natural nasal breathing, we obtained at least a sufficient nasal breathing allowing the patient to improve the chewing and that was the most important purpose the patient was waiting for. A paramedian forehead flap was raised using a plastic template and rotated over the graft and sutured with the lining flaps to cover the grafts entirely. To clear the nostril openings two nasal tubes were putted and sutured inside the new nose to leave the respiratory tract clear. Finally, the forehead flap was rotated over the graft and sutured with the lining flaps to cover the grafts entirely and the donor site was primarily closed and two nasal tubes were placed in the newly created nares and held in place for 6 months (Figure 2 (c, d, e)).

The transverse expansion was achieved by a tri-helix manually shaped so that it is perfectly adapted to this very narrow shape of the palate. This early removable maxillary expansion (ERME) could expand the maxillary basal bone arch width, nasal cavity width, maxillary alveolar bone arch width, and maxillary dental arch width. Secondary increases in the mandibular alveolar bone and dental arch widths would happen after ERME. To continue the expansion of this this narrow transverse dimension, the tri-helix has been replaced by a Hyrax expander chosen from the trade and fixed by two screws at the level of the palate. The hyrax is used at the same time as an anchoring means for the traction of the impacted incisors using elastomeric chains. The other teeth were subsequently pulled to an ideal position which allows the bonding of the brackets (Figure 2b).

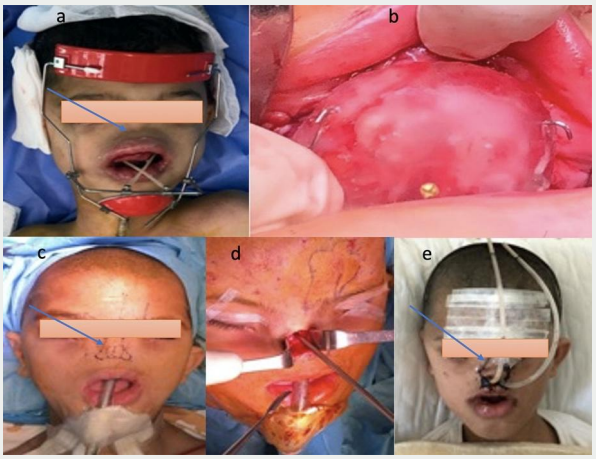

Figure 2. Facemask **(a)** supported by a resin splint screwed into the hard palate **(b)** and extraoral photographs showing the nasal reconstruction **(c, d, e)**. The arrow in figure **(a and c)** shows a complete absence of the external nose and in figure **(e)** shows the newly created nares with the two nasal tubes placed inside.

Follow-up and outcomes of interventions

Postoperative follow-up shows good esthetic and functional results of the new nose 3 years after surgically reconstruction (Figure 3a). Postoperative x-rays show the effectiveness of the orthodontic traction of impacted teeth and the nasal tube stability inside the new nose which allows efficient nasal breathing (Figure 3 (b, d)). The lateral cephalometric analysis and superimposition of before and after distraction showed the efficiency of the Delaire face mask for external distraction, since the ANB went from -11° to 1° (Figure 3c and Table 2). Postoperative 3D CT scan shows the effectiveness of the nasal reconstruction with nasal tube stability and orthodontic traction of impacted teeth. No internal nose restenosis, a slight external restenosis of a left nave was observed (Figure 3d) because of the loss of one nasal tube, which requires a reoperation to put it back. He is currently alive and is 13 years old and he continues his orthodontic treatment. An improvement in the quality of life was observed in this child's life in general with a positive psychosocial impact.

Table 2. Resumes pretreatment and posttreatment cephalometric examination of our patient.

Variable (in degrees)	Norms	Before	After
Sella Nasion Point A angle (SNA)	82±2°	73°	88°
Point A Nasion Point B angle (SNB)	2±2°	-11°	1°
Sella Nasion Point D angle (SND)	75°	79°	83°
Mandibular plane (GoGn) to cranial plane (SN) angle (GoGnSN)	32±5°	39°	39°
Upper incisor to Nasion Point A angle (ItoNA)	24±2°	14°	28°
Lower incisor to Nasion Point B angle (itoNB)	24±2°	21°	21°
Upper incisor to lower incisor angle (Itoi)	130±5°	151°	130°

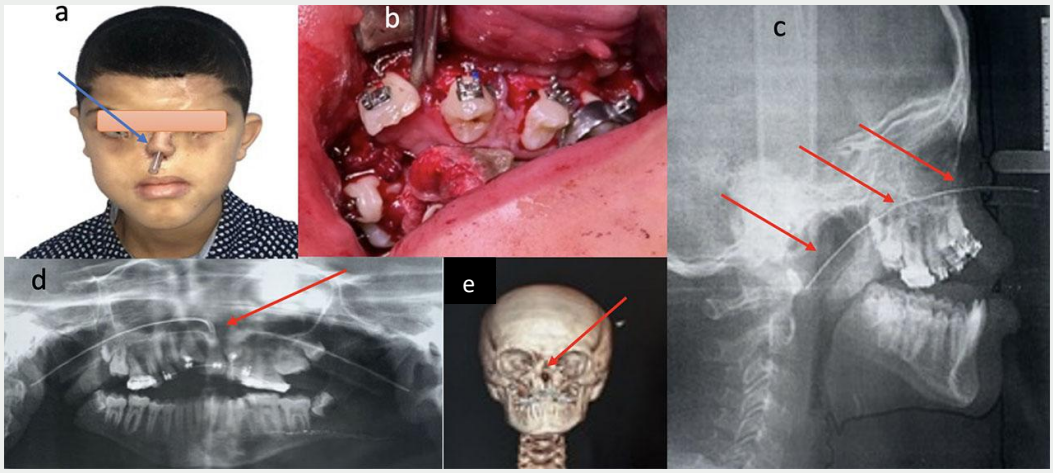

Figure 3. Postreatment extraoral **(a)**, and intraoral photographs **(b)**, and radiographs **(c, d)** and 3D CT scan **(e)**. The arrow in figure **(a)** shows a good esthetic and functional results of the new nose 3 years after surgically reconstruction and in figures **(c, d, e)** shows the effectiveness of this reconstruction.

Discussion

This case report highlights a very rare presentation of AS in orthodontic patient. The successful surgical management emphasizes the importance of considering interdisciplinary relationship between the orthodontist and a maxillofacial surgeon.

Because AS is exceptionally rare, this is the only case in Morocco among fewer than 30 cases described in literature worldwide with AS (7). Because AS is exceptionally rare, the pathogenesis of this disease has not been fully understood. It is postulated that lack of development of the nose results from medial failure and lateral nasal process growth, but it is also possible that overgrowth and premature fusion of the nasal medial process result in the formation of the atretic plate (2).

Whole-exome sequencing was employed to identify the genetic entity of this child led to the identification of de novo heterozygous missense mutations in the SMCHD1 gene. Heterozygous missense mutation was identified in this case and the variants were de novo, suggesting germline mutations in parental gametes. (6). Yang reported a heterozygous pathogenic variant, NM_015295, c.1025G > C; p. (Trp342Ser) of SMCHD1 was identified (5).

The reconstruction of AS is very complex and it should be performed only by a multidisciplinary team that includes oto-laryngology, plastic surgeons, and orthodontist. The arhinia reconstruction progress mainly consists of two parts: reconstruction of the nasal cavity and reconstruction of the external nose. The literature includes some reports of neonatal respiratory distress accompanying this disease (8, 10). Remarkably, our patient did not show any signs of respiratory distress except some difficulty in breathing and eating after surgery.

The second point to be managed in arhinia is the definitive correction of the malformation with functional and esthetic aims. An integrated approach should address all the issues of the disease, and we believe that the treatment objectives are to create a wide nasal cavity and an external nose. Finally, the ideal treatment should have the lowest possible morbidity and a low relapse rate (3). But clearly, our patient needed maxillary expansion and impacted tooth placement.

The most difficulty in the approach is the creation and maintenance of a nasal cavity. Because of the completely subverted local anatomy due to the reduced vertical dimensions of the palatal vault and ethmoid, the procedure is technically difficult to realize and quite risky. Moreover, there is a definite risk of entering the cranium while trying to create a sufficiently wide nasal cavity because the maxilla and palate are in close proximity to the cranial base (3). In addition, because restenosis of the new cavity with time, there is a definite relapse rate reported in the literature. Our patient showed a slight external restenosis of a nave on the left side because of the loss of one nasal tube, which requires a reoperation to put it back. Finally, some techniques require dental extractions to create space for the new cavity (10), but for our patients we don't need to extract any teeth.

In accordance with Brusati and Colletti (4) maxillary osteotomy allows one to address the maxillary vertical hypoplasia, which can normalize the labial-dental proportions and dental appearance simultaneously. But in our case, this is not sufficient because, there is a severe narrowness of the transverse dimension to the maxilla which contaminates the teeth in retention. For this reason, in our opinion it is essential to act on the transverse direction in parallel with the different therapeutic means to control the eruption of the impacted teeth.

The last treatment objective is the creation of an external nose. This is attained with a combination of standard techniques, including a forehead flap and bicortical graft. An acceptable morphology for the newly created external nose was obtained. This result was be stable over three years. One last controversy pertains to the optimal timing of treatment. We agree with the general statement reported by Brusati and Colletti (4).

Conclusions

A Moroccan male child, 7-year-old presented a complete absence of the external nose, nasal cavities, and other

characteristic signs of AS with difficulty in breathing and inability to feed due to airway obstruction. This diagnosis was genetically confirmed. A complex surgically reconstruction of this case was performed by a multidisciplinary team starting with a Le Fort II maxillary osteotomy with a bicortical approach under submental intubation. The effect of this osteotomy was maintained and potentiated by Facemask. The patient benefited from a parietal bone graft. The patient underwent an internal and external nasal reconstruction. Tri-helix and Hyrax used to expand maxilla and to ensure anchor for impacted teeth. The use of facemask therapy with a miniscrew potentiated the effect of osteotomy and stabilized the parietal bone graft which allowed nasal reconstruction and orthodontic management of unerupted teeth in this case.

References

1. Brusati R, Donati V, Marelli S, Ferrari M. Management of a case of arhinia. J Plast Reconstr Aesthet Surg 62:e206, 2009.
2. Thornburg LL, Christensen N, Laroia N, Pressman EK. Prenatal diagnosis of total arhinia associated with normal chromosomal analysis: a case report. J Reprod Med. 2009 Sep;54(9):579-82. PMID: 19947037.
3. Zhang MM, Hu YH, He W, Hu KK. Congenital arhinia: A rare case. Am J Case Rep. 2014;15:115-118.
4. Brusati R. and Colletti G. The role of maxillary osteotomy in the treatment of arhinia. J Oral Maxillofac Surg 70:e361-e368, 2012.
5. Yang JL, Gu H, Yuan ZZ, Xie XH, Yang YF, Tan ZP. Identification of a pathogenic SMCHD1 variant in a Chinese patient with bosma arhinia microphthalmia syndrome: a case report. BMC Med Genomics. 2024;17(1):136.
6. Gordon CT, Xue S, Yigit G, Filali H, Chen K, Rosin N, Yoshiura KI et al. De novo mutations in SMCHD1 cause Bosma arhinia microphthalmia syndrome and abrogate nasal development. Nat Genet. 2017;49(2):249-55.
7. Akkuzu G, Akkuzu B, Aydin E, Derbent M, Ozluoglu L. Congenital partial arhinia: a case report. J Med Case Rep. 2007;1:97.
8. Sohrabi C, Mathew G, Maria N, Kerwan A, Franchi T, Agha RA. The SCARE 2023 guideline: updating consensus Surgical CAse REport (SCARE) guidelines. Int J Surg Lond Engl. 2023;109(5):1136.
9. Kılıçoğlu H, Öğütlü NY, Uludağ CA. Evaluation of skeletal and dental effects of modified jasper jumper appliance and delaire face mask with pancherz analysis. Turk J Orthod. 2017;30(1):6-14.
10. Feledy JA, Goodman CM, Taylor T, Stal S, Smith B, Hollier L. Vertical facial distraction in the treatment of arhinia. Plast Reconstr Surg. 2004 Jun;113(7):2061-6.