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Bilateral Athelia: A Case Report

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Case Report | **Abstract:** | **DOI:**

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Background: Bilateral athelia, the congenital absence of both nipples and areolae, is an extremely rare condition. This case report documents the presentation, diagnosis and surgical management of bilateral athelia in a 22-year-old female from Bangladesh, highlighting the clinical implications, challenges and outcomes of reconstructive procedures. **Case Presentation:** The patient, born to non-consanguineous parents following an uneventful pregnancy and normal vaginal delivery, presented with bilateral athelia noted in the postnatal period. Despite initial consultations with local physicians, no diagnosis or treatment was provided. The patient's developmental milestones were normal and she experienced regular menarche at 13 years. At 21, she sought care at Bangladesh Specialized Hospital, Dhaka, for severely hypoplastic breasts and the absence of the NAC. Physical examination revealed well-developed pectoral muscles, small breast mounds, hypopigmentation, sparse hair, symbrachydactyly of the toes and myopia. Laboratory tests, including hormonal assays and chromosomal analysis, were normal. Ultrasound confirmed the absence of the NAC and the presence of glandular tissue substituted by adipose tissue. **Intervention:** Under general anesthesia, bilateral breast augmentation was performed using autologous fat transfer. Nipple-areola reconstruction was achieved using the skate flap technique and full-thickness skin grafting. Follow-up included a second session of fat grafting to enhance results and tattooing of the NAC for aesthetic improvement. **Results:** The patient reported high satisfaction with the aesthetic and functional outcomes. The postoperative course was uneventful, with no significant complications observed. The follow-up procedures significantly enhanced the aesthetic results, demonstrating the effectiveness of the chosen surgical techniques. **Conclusion:** This case highlights the successful management of bilateral athelia through comprehensive diagnostic evaluation and advanced reconstructive surgery. Early diagnosis, meticulous surgical planning and follow-up procedures are critical for achieving satisfactory outcomes in patients with rare congenital anomalies like athelia. Further research and case documentation are necessary to develop standardized management protocols and improve patient care.

Keywords: Bilateral Athelia.

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INTRODUCTION

Bilateral athelia, characterized by the congenital absence of both nipples and areolae, is an exceedingly rare congenital anomaly. Understanding this condition is crucial for improving diagnostic and management strategies, especially in regions with limited access to specialized healthcare. This case report documents a unique presentation of bilateral athelia in a Bangladeshi patient, emphasizing its clinical significance, challenges in diagnosis and broader implications for healthcare professionals[1]. Athelia is defined as the congenital absence of the nipple-areola complex, distinct from related conditions such as amastia, which involves the absence of breast tissue and hypoplasia, characterized by underdeveloped breast tissue. The absence of the nipple and areola can significantly impact physical appearance and psychological well-being, affecting body image and self-esteem and complicating breastfeeding in females[1]. While the global prevalence of athelia is not well-documented due to its rarity, it is often observed in association with other congenital anomalies or syndromes, such as Poland syndrome, limb-mammary syndrome and ectodermal dysplasias[2,3]. The etiology of athelia involves a complex interplay of genetic, developmental and environmental factors. Genetic mutations, such as those in the PTPRF gene, have been identified as significant causes, affecting the development of the nipple-areola complex. These mutations disrupt epithelial cell-cell contacts, peptide growth factor signaling and the canonical Wnt pathway, crucial for nipple development[4]. Developmental anomalies and syndromic associations further complicate the clinical picture, making early diagnosis and intervention essential[5]. Previous case reports and studies on athelia have highlighted

its diverse presentations and associations with other anomalies. One notable case reported a lethal syndrome of choanal atresia, athelia, renal tubulopathy and a family history of neck cysts, providing valuable insights into the genetic and developmental aspects of the condition[6]. Another study documented the phenotypic characteristics of patients with athelia and tooth agenesis, emphasizing the common deformities in hair, skin and sweat glands and identifying syndromes related to nipple deformity and their associated genes[7]. These studies underscore the importance of documenting and understanding the diverse manifestations of athelia to improve clinical outcomes. Despite these insights, significant knowledge gaps remain in the current literature. There is a need for more comprehensive studies to elucidate the full spectrum of genetic and environmental factors contributing to athelia. Further research is required to confirm the role of parathyroid hormone-related protein in nipple development and to explore potential therapeutic interventions[8]. Additionally, understanding the psychosocial impacts of athelia on affected individuals is crucial for providing holistic care. The physical, psychological and social impacts of athelia are profound, affecting the quality of life of individuals. The condition can lead to significant body image issues, particularly in societies where physical appearance is closely linked to self-esteem and social acceptance[9]. Moreover, the rarity of the condition poses challenges in diagnosis and management, as healthcare professionals may have limited experience with athelia. Early diagnosis and intervention are therefore critical to mitigate these impacts and provide appropriate psychosocial support and potential reconstructive options[10]. The importance of early diagnosis cannot be overstated. Early identification of athelia allows for timely interventions, such as genetic counseling, psychosocial support and reconstructive surgery, which can significantly improve the quality of life for affected individuals. Early diagnosis also facilitates the management of associated anomalies and syndromes, reducing the risk of complications and improving overall outcomes[11]. In regions with limited access to specialized healthcare, such as Bangladesh, raising awareness among healthcare providers and integrating athelia screening into routine pediatric assessments can enhance early detection and intervention efforts[12]. This case report aims to contribute to the existing literature on athelia by documenting a unique presentation of bilateral athelia in a Bangladeshi patient. By highlighting the clinical presentation, diagnosis and management of this case, we aim to raise awareness among healthcare professionals and emphasize the importance of early diagnosis and intervention.

CASE PRESENTATION

We present the case of a 22-year-old female from Bangladesh, who was born as the second child to non-consanguineous parents following an uneventful pregnancy and normal vaginal delivery. The postnatal period was marked by the early discovery of a rare congenital anomaly the absence of the nipple-areola complex (NAC) on both breasts. Initial attempts at seeking medical advice from local physicians proved fruitless, as none could provide an explanation or a treatment path for this deformity. Apart from the absence of the NAC, no other physical abnormalities were noted at birth.

The patient's developmental milestones were achieved timely, with menarche commencing at 13 years, characterized by a regular menstrual cycle. However, as she matured, concerns regarding breast development became pronounced. At the age of 21, she sought care at the plastic surgery department of Bangladesh Specialized Hospital, Dhaka, presenting with severely hypoplastic breasts and athelia.

Physical examination revealed a body weight of 48 kilograms and a stature of 5 feet 2 inches. The chest examination showed well-developed pectoral muscles, small breast mounds and a total absence of the NAC bilaterally. Additional findings included hypopigmentation on the abdominal and chest skin, sparse hair and symbrachydactyly affecting both the 3rd and 4th toes. The patient also reported myopia, for which she had been previously prescribed corrective lenses.

An extensive workup, including a complete blood count, biochemistry panel and hormonal assays encompassing progesterone, luteinizing hormone (LH), follicle-stimulating hormone (FSH), estradiol and testosterone levels, revealed no abnormalities. A chromosomal analysis confirmed a normal female karyotype of 46XX. Diagnostic imaging of the breasts was performed, with ultrasound findings detailed as follows: Ultrasonography of the breasts showed a moderate amount of glandular tissue substituted by adipose tissue and no abnormalities in the chest muscles. The patient's serum parathyroid hormone-related protein (PTHrP) levels were within normal limits.

Procedure

Under general anesthesia, bilateral breast augmentation was performed using autologous fat transfer. Fat was harvested from the abdomen, processed and then injected into the breast areas, mainly in the subcutaneous plane. Nipple-areola reconstruction was performed in the same session. Nipple projection was attained with the help of a skate flap and the areolae were reconstructed by full-thickness skin grafting. To enhance the aesthetic outcome, tattooing of the NAC was performed at a later date.

Since a significant amount of fat grafting is typically reabsorbed within the first few months, a second session of fat grafting was conducted four months after the initial surgery.

Overall, the patient's postoperative course was uneventful and she reported satisfaction with the aesthetic and functional outcomes of the reconstructive procedures. This case underscores the importance of early diagnosis and intervention and

highlights the effectiveness of modern reconstructive techniques in managing rare congenital anomalies such as bilateral athelia.

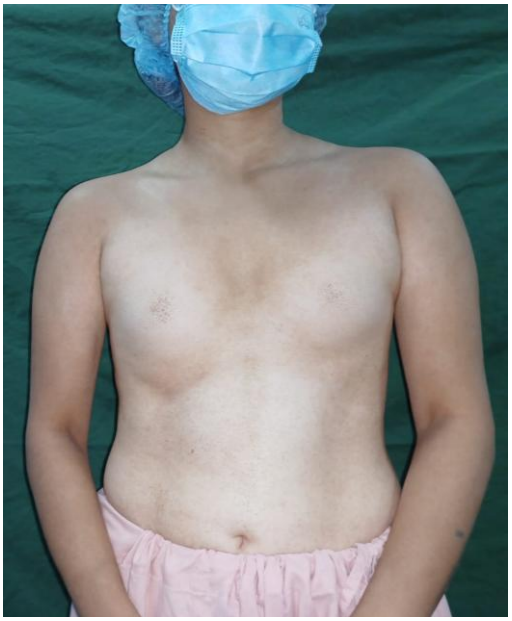

Figure 1: Athelia (absence of nipple and areola) in a 22 year-old female.

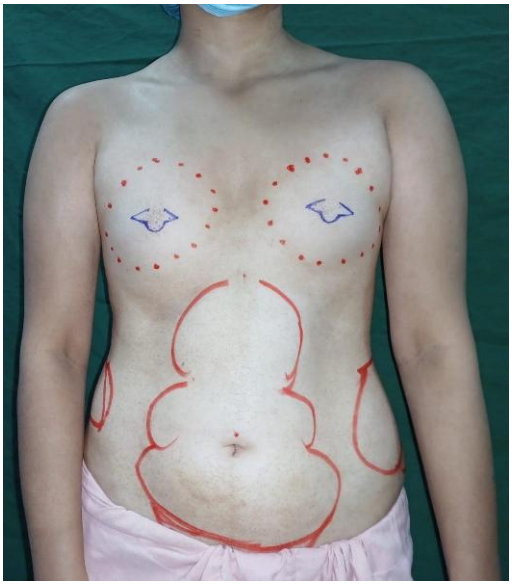

Figure 2: Preoperative markings for Nipple reconstruction by Skate flap (blue ink), Lipoinjection of breasts (dotted red ink) and Liposuction from abdomen and waist (red line).

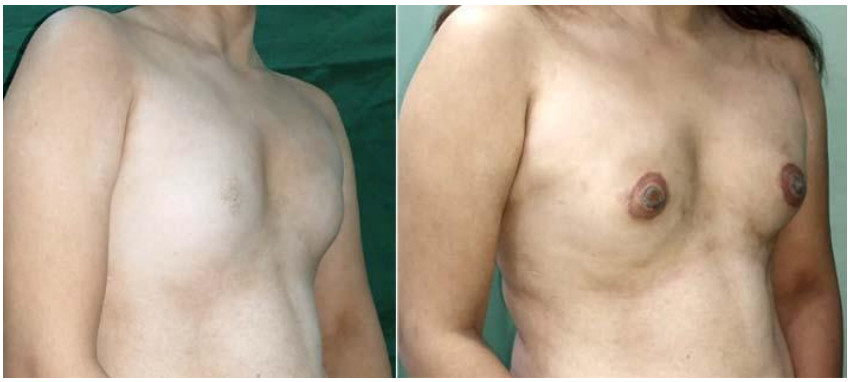

Figure 3: Before and after Nipple Areola Reconstruction and Lipoinjection of breasts

DISCUSSION

This case of bilateral athelia in a 22-year-old female from Bangladesh presents unique challenges and insights into the clinical management and understanding of this rare congenital anomaly. Athelia, characterized by the absence of the nipple-areola complex (NAC), is a rare condition with significant physical, psychological and social impacts on affected individuals. Previous case reports and studies have highlighted various etiological factors, including genetic mutations and developmental anomalies, which can contribute to the condition. For instance, a study by Borck et al. identified a homozygous frameshift mutation in the PTPRF gene, which disrupts the development of the NAC[2]. The clinical implications of athelia extend beyond the physical absence of the NAC. The condition can lead to significant psychological distress, impacting body image and self-esteem, particularly in societies where physical appearance is closely linked to social acceptance. Bouman et al. assessed the long-term psychosocial functioning of children with esophageal atresia, drawing parallels to the psychological impacts of congenital anomalies like athelia[13]. The need for early diagnosis and intervention is critical to mitigate these impacts and provide appropriate psychosocial support and potential reconstructive options. Gagnon highlighted the importance of comprehensive prenatal diagnostic workups, including ultrasound and other imaging techniques, to detect congenital anomalies early[14]. In this case, the patient's diagnostic workup included a complete physical examination, imaging and laboratory tests. The normal hormonal assays and chromosomal analysis results ruled out other conditions, providing a clear diagnosis of bilateral athelia. Diagnostic imaging, particularly ultrasound, confirmed the absence of the NAC and the presence of glandular tissue substituted by adipose tissue. This aligns with the findings of Gliozheni et al., who emphasized the role of ultrasound in diagnosing fetal anomalies[15]. The surgical intervention involved autologous fat transfer and nipple-areola reconstruction. Autologous fat transfer has been shown to be a safe and effective method for breast reconstruction, as demonstrated by Missana et al., who reported high patient satisfaction and good cosmetic outcomes[16]. The use of the skate flap and full-thickness skin grafting techniques for NAC reconstruction further improved the aesthetic results. Nakagawa et al. described similar techniques in their study, highlighting their effectiveness in achieving satisfactory nipple projection and areola pigmentation[17]. The outcomes of the surgical intervention in this case were positive, with the patient reporting high satisfaction with the aesthetic and functional results. However, the need for follow-up procedures, such as additional fat grafting sessions, was necessary to enhance the results and compensate for fat reabsorption. Sorotos et al. emphasized the importance of follow-up in their study on systematic fat transfer protocols for breast reconstruction, noting that multiple sessions can significantly improve long-term outcomes[18]. Despite the successful management of this case, several challenges and limitations were encountered. The rarity of bilateral athelia poses difficulties in diagnosis and management, as healthcare professionals may have limited experience with such cases. Goldman et al. discussed the challenges faced in autologous breast reconstruction, including the need for multiple procedures and potential donor site morbidity[19]. Additionally, the need for further research to generalize findings and develop standardized protocols for the management of athelia remains critical. Cho et al. highlighted the importance of addressing complications, such as NAC necrosis and developing techniques to mitigate these risks in high-risk patients[20]. In conclusion, this case report underscores the importance of early diagnosis, comprehensive management and the use of advanced surgical techniques in improving outcomes for patients with bilateral athelia. The successful reconstruction achieved in this case, coupled with high patient satisfaction, highlights the effectiveness of autologous fat transfer and nipple-areola reconstruction techniques. Continued research and case documentation are essential to enhance our understanding and management of this rare condition, ultimately improving the quality of life for affected individuals.

CONCLUSION:

In conclusion, this case of bilateral athelia in a young female from Bangladesh underscores the complexities and challenges associated with diagnosing and managing rare congenital anomalies. Through a comprehensive diagnostic workup and innovative surgical interventions, including autologous fat transfer and nipple-areola reconstruction, the patient achieved satisfactory aesthetic and functional outcomes. This case highlights the importance of early diagnosis, meticulous surgical planning and follow-up procedures to enhance patient satisfaction and long-term results. Further research and documentation of similar cases are essential to develop standardized protocols and improve clinical outcomes for patients with athelia.

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