Ref: Ro Med J. 2021;68(Suppl5)
DOI: 10.37897/RMJ.2021.S5.8

Preauricular tags and pits – a minor anomaly in newborns, and when further investigation is needed

Silvia-Nicoleta Radulescu[1], Vlad Dima[2], Simona Vladareanu[3,4]

[1]"Prof. Dr. Alexandru Trestioreanu" Institute of Oncology, Bucharest, Romania
[2]Department of Neonatology, Filantropia Clinical Hospital, Bucharest, Romania
[3]Elias University Emergency Hospital, Bucharest, Romania
[4]"Carol Davila" University of Medicine and Pharmacy, Bucharest, Romania

ABSTRACT

Preauricular skin tags represent a commonly seen minor congenital anomaly of the auricle that may occur either in isolation or together with other dysmorphic features or malformations as part of a syndrome. Even minor dysmorphic features can lead to a genetic diagnosis with far-reaching, systemic implications.

Neonatologists must be familiar with this common presentation and have good communication with the medical geneticist to identify a possible underlying genetic condition early and manage each case appropriately.

Ear abnormalities such as preauricular tags and pits may be associated with additional specific abnormalities such as hearing loss or renal malformations that need to be considered and further investigated.

This article aims to provide data and guidance for the management of neonates with preauricular tags or preauricular pits, a minor congenital anomaly of the ear, where early recognition of syndromes based on minor anomalies is ideal.

Keywords: preauricular tags, minor congenital anomalies, renal malformations, hearing loss

INTRODUCTION

A birth defect is a structural anomaly present at birth that may be caused by genetic factors and/or environmental exposures, although there are many cases with unknown etiologies (1).

The study of these structural defects, dysmorphology, aims to interpret patterns of human growth and morphological abnormalities that fall outside the normal developmental range, either by descriptive observation or measurement (2,3,4). These include malformation, disruption, deformation, and dysplasia, the four major types of problems in morphogenesis (2,5). The diagnosis of the dysmorphic syndrome is part of the typical clinical genetics practice (6). Children with a congenital defect and/or a dysmorphic syndrome account for 2-3% of live births (2).

Major congenital defects are defined as anomalies that have medical, surgical, or cosmetic significance (1). Major birth defects occur in 1% of newborns and are more common in stillbirths and spontaneous miscarriages (1,7). Fortunately, thanks to advances in prenatal diagnosis, most severe anomalies are detected before birth (7).

Minor structural anomalies are more common than major malformations and occur in 3% of newborns (1,7). Common or normal variants have a prevalence above 4% in the general population and minor anomalies have a prevalence </=4% in the general population (8).

Neonatologists frequently encounter newborns with dysmorphic features. They are often the first physicians to evaluate these appearances (9). Auricular anomalies are frequently implicated in genetic pathology, and even minor variants may be an important indicator for the detection of some genetic syndromes (5,8). Neonatologists need to be familiar with this common finding so that they can perform

Corresponding author:
Vlad Dima
E-mail: dima.vlad@yahoo.com

Article History:
Received: 4 November 2021
Accepted: 29 November 2021

further testing for occult malformations as needed and collaborate with the medical geneticist to perform diagnostic testing thoroughly and counsel families appropriately (9).

This article addresses the syndromic aspects of preauricular skin tags and preauricular pits, a minor anomaly of the ear. We describe the main potential associations of this presentation and give a brief outlook on when these cases should be further investigated.

ANATOMY AND EMBRYOLOGY OF THE EAR

Congenital anomalies of the external ear arise from defects during the embryonic development of the fetal auricular cartilage (10). Auricular anomalies are highly variable structural defects that can range in severity from minor to major anomalies (8,10).

The anatomy of the ear is complex. The external ear, also known as the auricle or pinna, develops in the 6th to 12th weeks of gestation and is derived from the six auricular hillocks of the first and second branchial arches (8,10,11,12).

Preauricular tags are small non-cartilaginous skin-colored protrusions that can be found anterior to the insertion of the ear usually hairless and containing no striated muscle (9,11). Preauricular pits are small skin openings of variable locations. Preauricular pits are also known as preauricular cysts, fissures, or sinuses (9,13). Supernumerary development of the first 3 hillocks of the first branchial arch may be the cause of the preauricular tags (14).

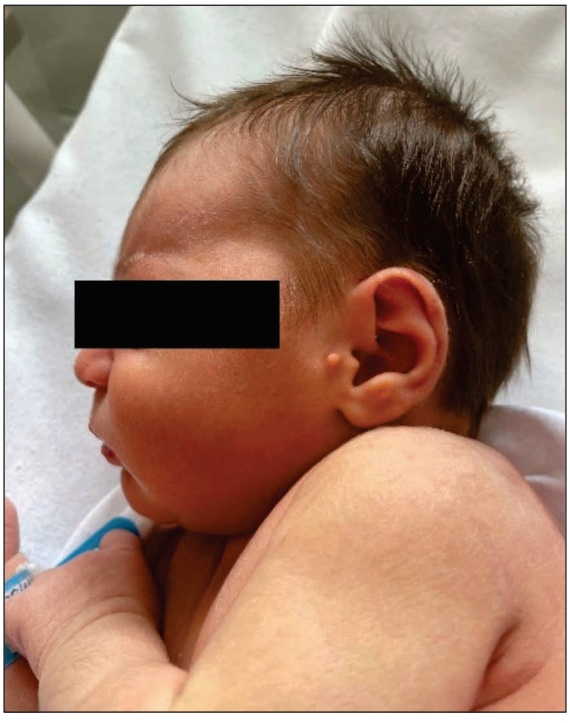

FIGURE 1. Newborn with preauricular skin tag (personal collection Silvia-Nicoleta Rădulescu)

ASSOCIATED MALFORMATIONS AND GENETIC SYNDROMES

Preauricular tags and pits are minor congenital anomalies that are highly variable and are common findings on routine neonatal examination, occurring in 0.5-1% of newborns (8,9,10). They may occur either in isolation or in a characteristic combination or pattern that may affect one or more organ systems as part of a syndrome (1,9). In addition, many genetic syndromes are recognized by the pattern of minor abnormalities noted on clinical examination (9).

Approximately 30% of congenital ear malformations are associated with additional abnormalities within a genetic syndrome (13). Additional abnormalities are present in approximately 20% of newborns with ear tags or pits. Therefore, a careful general assessment is required before considering preauricular tags or pits as an isolated trait (8).

Newborns with preauricular tags or pits are at increased risk for permanent hearing impairment (15). Infants with isolated preauricular tags and pits may also be more likely to have eustachian tube dysfunction (16).

When it comes to isolated preauricular tags, in 2006 Yongjia Yang identified the locus for isolated autosomal dominant accessory auricular anomaly (ADAAA). The locus identified was 9.84 cM interval between the markers D14S283 and D14S297. This study assigned an isolated ADAAA locus to 14q11.2–q12 (17).

Syndromic diagnoses to be considered include Brachio- oto-renal (BOR) syndrome, oculo-auriculo-vertebral dysplasia (OAV or Goldenhar syndrome), mandibulofacial dysostosis (Treacher-Collins syndrome), craniofacial dysostosis (Crouzon syndrome, Apert syndrome), oto-cervical dysostosis (Klippel-Feil syndrome, Wildervanck syndrome), otoskeletal dysostosis (osteogenesis imperfecta, Albers-Schönberg syndrome), and various chromosomal abnormalities (5,8,9,13).

De novo ear abnormalities may also be associated with injury during pregnancy such as infections, chemical agents, malnutrition, irradiation, rhesus incompatibility, hypoxia, air pressure fluctuations, and noise exposure (13). Exogenous factors are thought to contribute to malformations of the external ear by 10% (13).

MANAGEMENT OF CASES

In a newborn with preauricular tags and/or pits, it is important to perform a complete clinical examination, including growth parameters, and to look for other possible associated dysmorphic elements or malformations that may indicate an underlying genetic syndrome (7,9). A closer look at the family

history with an emphasis on ear abnormalities, deafness, renal abnormalities, and dysmorphology is also required (8).

Good collaboration within the team of neonatologists and geneticists is important to promptly investigate these cases further and make a correct diagnosis, ultimately confirming a genetic etiology as soon as possible, identifying associated defects early, providing better treatment options and quality of life, and ensuring proper family genetic counselling regarding the risk of recurrence and impact on other family members at risk (8).

HEARING EVALUATION

Approximately 17% of newborns with isolated tags or pits have conductive and/or sensorineural hearing impairment (8) All infants with a preauricular tag or pit should be evaluated audiologically because abnormalities of the external ear may be associated with middle or inner ear abnormalities and progressive hearing loss (9,14)

Transient evoked otoacoustic emissions (TEOAEs) effectively investigate hearing in infants with preauricular skin tags or ear pits (15). Children with isolated preauricular tags or pits without risk factors for hearing loss may not need hearing assessment outside of regular hearing screening (16).

RENAL ULTRASOUND SCAN

The association between preauricular ear tags and pits and renal anomalies has also been studied (9) In children with isolated preauricular ear tags, the incidence of urinary tract abnormalities on renal ultrasound is 3% to 8%. In the general pediatric population, structural abnormalities of the kidney occur in 1-3% of live births (18).

Renal US should only be performed in cases with bilateral ear tags and/or pits or when accompanied by one or more of the following: other malformations or dysmorphic features, hearing loss, ear anomalies or renal malformations in the family history, maternal diabetes, or teratogen exposure. If these findings are not present, no further investigation is indicated (8,9,10,19).

SURGICAL TREATMENT

Sometimes the tags are purely skin only, but usually the tag contains a long tail of cartilage extending into the cheek. It is not possible to tell whether the procedure could be done simply or whether it would require a general anesthetic until a surgeon evaluates the skin tag. If a general anesthetic is required, then it would usually be recommended that the procedure is left until your child is older.

If the base of the tag is relatively narrow, and the cartilage component relatively minor, tags can be removed soon after birth using a clip device which requires no anesthetic. The clip cuts off the blood supply to the tag, and after a few days, the additional tissues shrivel and separate. Tags with a substantial cartilage core are best treated by excision of the skin tag and cartilage spindle under general anesthetic preferably when baby is over the age of 2 or 3 years (20).

A lot of treatment options exist for skin tag in older children and in adult patients, such as cryosurgery, surgical excision, and electrosurgery, while ligature or excision are preferred for the newborns (21).

LOCAL INFECTIONS

It is generally accepted that most sinuses remain asymptomatic. In cases with sinus discharge, the likelihood of further episodes of discharge is also unclear. A small number will become repeatedly infected with local inflammation around the associated subcutaneous cyst and purulent discharge from the sinus. Despite careful surgical excision, recurrences are common – especially with infected lesions that required incision and drainage (22).

Some infected sinuses will cause chronic preauricular abscesses. This is thought to occur from rupture of the anterior wall of the preauricular cyst with spillage of the infected debris into the subcutaneous fat tissue. Intense chronic inflammation and foreign body reaction will appear alongside with fat necrosis and thinning and erosion of the overlying skin (23). Some authors recommended treating such chronic preauricular abscesses by complete resection of the preauricular sinus/cyst, the abscess wall and its overlying skin. This radical approach produces a large tissue defect, often requiring reconstruction with local flaps. The cosmetic results are less than optimal, but this is an efficient approach (24).

CONCLUSIONS

Preauricular tags and pits, even if they are minor anomalies of the ear, may be an important indicator for additional evaluation for other occult malformations that together constitute a genetic syndrome. This population should be carefully evaluated for other dysmorphic features or malformations that may indicate an underlying genetic syndrome.

Therefore, in these cases, it is crucial to have a strong interdisciplinary team that seeks to thoroughly evaluate the newborns with a suspected genetic disorder and select the appropriate tests to

confirm a genetic etiology with possible systemic and familial implications.

Hearing examination is required in all newborns because abnormalities of the external ear may be associated with hearing loss.

In view of the urinary tract malformations often associated with external ear anomalies, renal US is indicated only in selected cases. Infants must be evaluated if preauricular tags or pits are associated with other malformations or dysmorphic features, a family history of hearing loss or ear abnormalities, or a history of gestational diabetes or teratogen exposure, or in cases with a bilateral presentation.

Conflict of interest: none declared
Financial support: none declared

REFERENCES

1. Bacino CA. Birth defects: Epidemiology, types, and patterns. UpToDate. Available at: https://www.uptodate.com/contents/birth-defects-epidemiology-types-and-patterns.
2. Smigiel R, Demkow U. Next Generation Sequencing in Dysmorphology. In: Clinical Applications for Next-Generation Sequencing. Elsevier; 2016:137-51.
3. Sanchez-Lara PA. Clinical and Genomic Approaches for the Diagnosis of Craniofacial Disorders. *Curr Top Dev Biol.* 2015;115:543-59.
4. Ruíz VDC, Hernández RDU, Rosa GFZ. Genética clínica. *Manual Moderno*. 2019.
5. Graham JM, Sanchez-Lara PA. Smith's recognizable patterns of human deformation. 4th ed. Philadelphia, PA: Elsevier - Health Sciences Division; 2015.
6. Godbole K. Molecular dysmorphology. In: Clinical Molecular Medicine. Elsevier; 2020:89-105.
7. Johnson KA, Catanzarite V. The Term Newborn: Postnatal Screening and Testing. *Clin Perinatol.* 2021 Aug;48(3):555-572.
8. Firth HV, Hurst JA. Oxford Desk Reference: Clinical Genetics and Genomics. Oxford University Press, 2017.
9. Jones KL, Adam MP. Evaluation and diagnosis of the dysmorphic infant. *Clin Perinatol.* 2015 Jun;42(2):243-61, vii-viii.
10. Liaw J, Patel VA, Carr MM. Congenital anomalies of the external ear. *Operative Techniques in Otolaryngology-Head and Neck Surgery.* 2017;28(2):72-76.
11. Hunter A, Frias JL, Gillessen-Kaesbach G, Hughes H, Jones KL, Wilson L. Elements of morphology: standard terminology for the ear. *Am J Med Genet A.* 2009 Jan;149A(1):40-60.
12. Clark RD, Curry CJ. Genetic consultations in the newborn. Oxford University Press, 2019.
13. Bartel-Friedrich S, Wulke C. Classification and diagnosis of ear malformations. GMS Curr Top Otorhinolaryngol Head Neck Surg. 2007;6:Doc05.
14. Vlădăreanu S, Boț M, Berceanu C, Mehedințu C, Popescu S. The Neonate with Minor Dysmorphisms. In: Congenital Anomalies-From the Embryo to the Neonate. *IntechOpen,* 2017.
15. Roth DA, Hildesheimer M, Bardenstein S, Goidel D, Reichman B, Maayan-Metzger A, Kuint J. Preauricular skin tags and ear pits are associated with permanent hearing impairment in newborns. *Pediatrics.* 2008 Oct;122(4):e884-90.
16. Wu GT, Devine C, Xu A, Geelan-Hansen K, Anne S. Is routine audiometric testing necessary for children with isolated preauricular lesions? *Int J Pediatr Otorhinolaryngol.* 2017 Feb;93:68-70.
17. Yang Y, Guo J, Liu Z, Tang S, Li N, Yang M, et al. A locus for autosomal dominant accessory auricular anomaly maps to 14q11.2-q12. *Hum Genet.* 2006 Aug;120(1):144-7.
18. Izzedine H, Tankere F, Launay-Vacher V, Deray G. Ear and kidney syndromes: molecular versus clinical approach. *Kidney Int.* 2004 Feb;65(2):369-85.
19. Wang RY, Earl DL, Ruder RO, Graham JM Jr. Syndromic ear anomalies and renal ultrasounds. *Pediatrics.* 2001 Aug;108(2):E32.
20. Schonauer F, Guastafierro A, Grasso E, Izzo S, Nicoletti GF, D'Andrea F. Ligaclip for Preauricular Skin Tags in the Newborn. *Eur J Pediatr Surg.* 2021 Jun;31(3):273-275.
21. Görgülü T, Torun M, Güler R, Olgun A, Kargi E. Fast and Painless Skin Tag Excision with Ethyl Chloride. *Aesthetic Plast Surg.* 2015 Aug;39(4):644-5.
22. Rataiczak H, Lavin J, Levy M, Bedwell J, Preciado D, Reilly BK. Association of Recurrence of Infected Congenital Preauricular Cysts Following Incision and Drainage vs Fine-Needle Aspiration or Antibiotic Treatment: A Retrospective Review of Treatment Options. *JAMA* Otolaryngol Head Neck Surg. 2017 Feb 1;143(2):131-134.
23. Kim WJ, Lee YM, Kim DH, Choe S, Lee D, Park SY, Kang M, Cho HJ, Heo KW. Causes and prevention of revision surgery for preauricular sinus: A histopathological analysis. *Int J Pediatr Otorhinolaryngol.* 2019 Jan;116:199-203.
24. O'Brien JX, Ashton MW. Relationship of the temporofrontal rami of the facial nerve to the fascial layers of the temporal region. *Ann Plast Surg.* 2012 Jun;68(6):547-8.