Medical Science

To Cite:
Anioła A, Ważniewicz S, Saniuk J. Pilomatrixoma in childhood-diagnosis and treatment. A review of literature. *Medical Science* 2025; 29: e37ms3540
doi: https://doi.org/10.54905/disssi.v29i156.e37ms3540

Authors' Affiliation:
[1]University Clinical Hospital in Poznań, Przybyszewskiego 49, 60-355 Poznań, Poland
[2]Voivodeship Hospital in Poznań, Juraszów 7/19, 60-479 Poznań, Poland

[*]**Corresponding Author**
University Clinical Hospital in Poznań, Przybyszewskiego 49, 60-355 Poznań,
Poland
Email: sandrawazniewicz@gmail.com

Peer-Review History
Received: 29 October 2024
Reviewed & Revised: 02/November/2024 to 10/February/2025
Accepted: 13 February 2025
Published: 18 February 2025

Peer-review Method
External peer-review was done through double-blind method.

Medical Science
pISSN 2321–7359; eISSN 2321–7367

DISCOVERY SCIENTIFIC SOCIETY

Pilomatrixoma in childhood-diagnosis and treatment. A review of literature

Aleksandra Anioła[1], **Sandra Ważniewicz**[1*], **Jagoda Saniuk**[2]

ABSTRACT

Pilomatrixoma, a benign skin neoplasm growing from hair matrix cells, also known as calcifying epithelioma, is children's second most common benign skin tumor. Researchers reported two peaks in the incidence of the lesion, the first under the age of one and the second between the ages of 5 and 11. More often, the lesion appears in the female gender. Although the lesion is most often asymptomatic, proper diagnosis is essential. Histopathological examination confirms the diagnosis of pilomatrixoma. It is also crucial to exclude other skin lesions that may mimic pilomatrixoma, such as a dermoid or sebaceous cyst. The first choice of treatment is surgery; due to possible recurrence, it is necessary to excise the lesion with an adequate margin. The paper reviews the literature on pilomatrixoma in the pediatric population.

Keywords: Pilomatrixoma, children, Malherbe's calcifying epithelioma, beta-catenin, nodular lesion

1. INTRODUCTION

Pilomatrixoma is a benign skin neoplasm arising from hair follicle matrix cells in the dermis or subcutaneous tissue. Otherwise, it can also be known as Malherbe's calcifying epithelioma. It represents the second most common benign skin neoplasm in children (Kose et al., 2014). In children and adolescents, lesions mainly occur in the first twenty years of life. Lesions are usually described as single nodules on the head, neck, and upper extremities. Some patients have multiple lesions. However, this is often associated with the presence of a genetic syndrome (Laffargue et al., 2019). In addition, other forms of the lesion can present with rapid growth and lead to ulceration or perforation. Very rarely, giant pilomatrixomas, defined as lesions over 4 cm in diameter, are described (Tavares et al., 2022).

Objective

The purpose of the following paper is to review the literature on the occurrence of pilomatrixomas in the pediatric population and the diagnosis and treatment of the lesion.

2. METHODOLOGY

This work includes a review of the literature on the incidence of pilomatrixoma among children. We searched databases such as PubMed and Google Scholar for this review paper. We used keywords such as pilomatrixoma in childhood, pilomatrixoma in adolescence, calcifying epithelioma, diagnosis of pilomatrixoma in children, and treatment of pilomatrixoma in children. We found 47 published articles with the keyword "pilomatrixoma in childhood". This review paper included case reports of pilomatrixoma in children, review papers, and retrospective studies. We excluded papers older than 15 years, studies on the adult population, and papers with only abstracts available. We selected thirteen publications from January 2009 to June 2024.

3. RESULTS AND DISCUSSION

Epidemiology

Pilomatrixoma most often occurs in children and young adults. The highest percentage of diagnosed lesions occurs in the first two decades of life. Most researchers indicate that pilomatrixoma is more common in female patients. The incidence is not clearly defined (Jones et al., 2018). In an analysis of 171 cases of pilomatrixoma in children, Fu et al., (2024) described that the incidence had two peak points, under one year of age and between 5 and 11 years of age. In contrast, they described the fewest cases between the ages of 2 and 4 and over 12 (Fu et al., 2024). In the pediatric population, pilomatrixoma is the second most common benign skin tumor (Pelizzari et al., 2021).

Etiology

The etiology of pilomatrixoma is unknown. However, cases of lesions associated with genetic syndromes have been described. These include Turner syndrome, constitutional mismatch repair deficiency (CMMR-D), myotonic dystrophy, Kabuki syndrome, Stickler syndrome, or Rubinstein-Taybi syndrome. In such cases, several lesions are more common (Jones et al., 2018; Laffargue et al., 2019). In addition, the researchers described the presence of mutations in the CTNNB1 gene, which encodes beta-catenin. This protein is responsible for signaling in the Wnt pathway, which affects proliferation, cell differentiation into hair follicles, and cell adhesion. (Wachter-Giner et al., 2009; Jones et al., 2018).

Histopathology

Histopathologically, there are four stages of pilomatrixoma described as early, fully developed, early regressive, and late regressive (Jones et al., 2018). The histopathological picture includes circularly arranged cells. Basaloid basophilic cells are located peripherally and enucleated; eosinophilic "shadow cells" are located centrally. Basaloid cells have a small amount of cytoplasm and mitotic figures. Eosinophilic cells are without nuclei. In most cases, pathomorphologists observe calcification in the central areas. As the lesion increases, the number of basophilic cells decreases, and the number of "shadow cells" increases (Jones et al., 2018; Siadati et al., 2022).

Clinical presentation

Pilomatrixoma most often manifests as an asymptomatic subcutaneous nodule with slow growth. The skin around the lesion may be red or bluish, sometimes with accompanying inflammatory skin changes (Alkatan et al., 2021; Yang et al., 2023). Most often, nodules typically present with sluggish growth (Kose et al., 2014). The size of lesions usually ranges from 5mm to 20mm (Yang et al., 2023). Some papers describe lesions up to 20 cm in diameter (Jones et al., 2018).

Less commonly, some patients may develop ulceration, skin perforations, or discharge (Carcassola et al., 2022). Extremely rarely, transformation to a malignant lesion can occur (Brannigan et al., 2017). These lesions seldom recur (Siadati et al., 2022). Most lesions appear on the head, neck, upper extremities, and trunk (Laffargue et al., 2019). On the head skin, lesions often localize in the periorbital area, especially on the upper eyelids and eyebrows (Alkatan et al., 2021). Lesions are less common on the lower extremities (Jones et al., 2018).

Diagnostics

Diagnosis includes medical history and physical examination, as well as additional tests. Diagnosis by the clinical picture is complex (Siadati et al., 2022). During a physical exam, a "tent sign" can be helpful. It involves stretching the skin over the nodule, revealing numerous facets. In addition, a "teeter-totter" sign may be beneficial, in which pressure on one edge of the lesion causes the opposite edge to protrude (Alkatan et al., 2021). Researchers report the use of fine-needle aspiration biopsy for the diagnosis of pilomatrixoma. The disadvantage of this method is that it provides a correct diagnosis in only 40% of cases.

The aspirate rarely succeeds in observing both basaloid and eosinophilic cells, especially when the sample is taken only from the periphery of the lesion early. Therefore, fine-needle aspiration biopsy is rarely helpful for the diagnosis of pilomatrixoma (Jones et al., 2018). Another diagnostic method is dermatoscopic examination. On dermatoscopy, pilomatrixoma presents as a well-demarcated nodule with blue or blue-red coloration, with streaks and telangiectasias present (Wolff et al., 2014). Additional tests to expand the diagnosis include ultrasound, computed tomography (CT), positron emission tomography (PET), and magnetic resonance imaging (MRI) (Tavares et al., 2022).

There are five types of ultrasound images using the Solivetti classification. Type 1 describes a fully calcified lesion with a hypoechoic peripheral areola with the least vascularization. Type 2 includes solid, hypoechoic lesions with small calcifications and a hypoechoic halo surrounding peripheral vascularization. Type 3 are poorly demarcated lesions with anechoic areas and calcifications, no hypoechoic halo, and the lesions can be either non-vascularized or peripherally vascularized. Type 4 lesions are pseudocystic, without an acoustic shadow at the periphery.

In contrast, in type 5, irregular, pseudotumorous, solid lesions with the most significant internal and peripheral vascularization are observed (Pelizzari et al., 2021). CT scans most often image pilomatrixoma as a well-demarcated lesion with calcifications (Jones et al., 2018). This examination provides a good picture of the depth of the lesion; however, due to the high amount of radiation, it is not commonly recommended in children (Yang et al., 2023). On MRI, pilomatrixoma shows a signal of intermediate intensity at T1 and high intensity at T2 (Jones et al., 2018). However, MRI images of pilomatrixoma are not characteristic; therefore, this imaging has limited value in diagnosing the lesion (Yang et al., 2023).

On PET, the lesions show metabolic activity and calcification (Jones et al., 2018). The physician gives a final diagnosis based on histopathological examination (Kose et al., 2014). Table 1 summarizes the diagnostic methods. Differential diagnosis is essential, and several diseases require consideration. Pilomatrixoma can be confused with a sebaceous cyst or dermoid cyst. In addition, an infantile hemangioma, which localizes in the subcutaneous tissue and may be bluish, should be ruled out. For lesions localized on the eyelid, one should also keep in mind chalazion, which is a painless nodule (Yang et al., 2023).

Table 1 Summary of pilomatrixoma diagnosis.

Diagnostic Method	Result of the examination
Tent sign and teeter totter sign	Tent sign- multiple facets when the skin is stretched over the nodule. Teeter totter sign- protrusion of one edge when the opposite edge is pressed
Histopathological examination	Concentrically arranged basophilic cells in the periphery and non-nucleated eosinophilic cells in the center (“shadow cells”)
Ultrasound	Lesions with various degrees of calcification, often with the presence of a hypoechogenic halo at the periphery
CT	Limited lesions with the presence of calcifications
MRI	Lesions with the presence of calcification. In the T1 sequence with intermediate signal intensity, in the T2 with high signal intensity
PET/CT	Metabolically active lesions with calcifications

Treatment

The literature does not describe spontaneous regression of pilomatrixoma, so surgical treatment is necessary. Researchers recommend complete resection of the lesion, including margins. Failure to perform a complete resection may result in the recurrence of the lesion (Jones et al., 2018). The authors define the recurrence rate as 0 to 3% (Kose et al., 2014). Due to its frequent location on the head and face, the minor possible incision is essential for a pleasing aesthetic result (Brannigan et al., 2017). Possible complications following the procedure include the occurrence of superinfection or wound dehiscence, hematoma formation, or hypertrophic scarring (Laffargue et al., 2019). It is scarce for transformation to a malignant lesion, which can yield organ metastases. In this case, radiation and chemotherapy are applicable in addition to surgical treatment (Kose et al., 2014).

4. CONCLUSIONS

Pilomatrixoma is a benign neoplasm from the hair matrix cells. It can occur at any age but is most often diagnosed in the first two decades of life. In children, pilomatrixoma is the second most common benign skin neoplasm. The etiology is still not fully understood. Researchers point to the possible involvement of mutations in the gene encoding beta-catenin. Symptoms of pilomatrixoma in children most often include the appearance of a painless, mobile nodule, usually located in the head and neck region. The basis of diagnosis is histopathological examination, which allows the diagnosis of pilomatrixoma. Additional tests such as ultrasound, CT, MRI, and PET can help in the differential diagnosis of the lesion. The physician should exclude the presence of a dermoid cyst, sebaceous cyst, or hemangioma. Treatment relies on surgical excision of the lesion with margins to prevent recurrence, so there is no recurrence.

Authors' Contribution

Sandra Ważniewicz: Conceptualization, writing- rough preparation, investigation, methodology, project administration, writing - review and editing

Aleksandra Anioła: Formal analysis, supervision, visualization, data curation, writing- rough preparation, writing - review and editing

Jagoda Saniuk: Methodology, data curation, resources, formal analysis

All authors have read and agreed to the published version of the manuscript.

Acknowledgments

No acknowledgments.

Ethical approval

Not applicable.

Informed consent

Not applicable.

Funding

This study has not received any external funding.

Conflict of interest

The authors declare that there is no conflict of interests.

Data and materials availability

All data sets collected during this study are available upon reasonable request from the corresponding author.

REFERENCES

1. Alkatan HM, Alhothali W, Alnuhayer O, Alessa D. Pilomatrixoma in a child mimicking a ruptured epidermal cyst clinically and histopathologically: Case report. Int J Surg Case Rep 2021; 84:106068. doi: 10.1016/j.ijscr.2021.106068

2. Brannigan K, Touil LL, Fattah AY. Multiple pilomatrixomas in children: the need for long-term follow-up and aesthetically sensitive incisions. Clin Exp Dermatol 2017; 42(4):400-402. doi: 10.1111/ced.13074
3. Carcassola MV, De-Campos WG, Lemos CA, Milani BA, Psciolaro RL, Ceccheti MM. Pilomatrixoma in the cheek: a clinical case report. Autops Case Rep 2022; 12:e2021387. doi: 10.4322/acr.2021.387
4. Fu H, Shen C, Wu B, Wang T, Wu J, Li Y, Ding J, Gao J. Clinical and Pathological Features of Pilomatrixoma in Children: A Retrospective Study. Dermatology 2024; 240(4):543-552. doi: 10.1159/000538802
5. Jones CD, Ho W, Robertson BF, Gunn E, Morley S. Pilomatrixoma: A Comprehensive Review of the Literature. Am J Dermatopathol 2018; 40(9):631-641. doi: 10.1097/DAD.0000000000001118
6. Kose D, Ciftci I, Harmankaya I, Ugras S, Caliskan U, Koksal Y. Pilomatrixoma in childhood. J Cancer Res Ther 2014; 10(3):549-551. doi: 10.4103/0973-1482.137918
7. Laffargue JA, Stefano PC, Vivoda JL, Yarza ML, Bellelli AG, Castro MN, Torres Huamani AN, Apa SN, Navacchia D, Centeno M, Cervini AB, Grees SA. Pilomatrixomas in children: Report of 149 cases. A retrospective study at two children's hospitals. Pilomatrixomas en niños: a propósito de 149 casos. Estudio retrospectivo en dos centros pediátricos. Arch Argent Pediatr 2019; 117(5):340-343. doi: 10.5546/aap.2019.eng.340
8. Pelizzari M, Giovo ME, Innocente N, Pérez R. Ultrasound findings in 156 children with 169 pilomatricomas. Pediatr Radiol 2021; 51(11):2038-2046. doi: 10.1007/s00247-021-05124-6
9. Siadati S, Campbell AA, McCulley T, Eberhart CG. Clinicopathological Features of 19 Eyelid Pilomatrixomas. Ocul Oncol Pathol 2022; 8(1):30-34. doi: 10.1159/000520219
10. Tavares AT, Neiva-Sousa M, Semedo C, Martins M, Gomes P. Pilomatrixoma Recurring as Giant Form. Cureus 2022; 14(1):e21308. doi: 10.7759/cureus.21308
11. Wachter-Giner T, Bieber I, Warmuth-Metz M, Bröcker EB, Hamm H. Multiple pilomatricomas and gliomatosis cerebri--a new association? Pediatr Dermatol 2009; 26(1):75-78. doi: 10.1111/j.1525-1470.2008.00827.x
12. Wolff CR, Brehmer F, Lockmann A, Hofmann L, Brauns B, Schön MP, Emmert S, Haenssle HA. Rapidly growing blue-red nodule on the cheek of a 4-year-old boy. Pilomatricoma. J Dtsch Dermatol Ges 2014; 12(5):432-434. doi: 10.1111/ddg.12281
13. Yang J, Zhao Y, Zhang H, Wang J. Periocular pilomatrixoma in childhood: Clinical feature and differential diagnosis. J Fr Ophtalmol 2023; 46(5):495-500. doi: 10.1016/j.jfo.2022.10.009