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Xanthoma in the external acoustic meatus

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ABSTRACT

Objectives: a unique case of a xanthoma in the external ear canal of a pediatric patient is presented in this case report. We describe and resume the current literature on xanthomas in the head and neck area.
Methods: clinical and intraoperative findings are reported and the presumed mechanisms for the emergence of xanthomas are discussed. We furthermore described the pathologic and immunohistochemical characteristics.
Results: xanthomas are mostly seen in patients with lipid metabolism disorders or hyperlipidemia. However, they can be present in normolipemic patients and are also associated with hematologic disease. Literature on the existence of xanthomata in the head and neck area is rare and there are no case reports in the pediatric population to the best of our knowledge. Besides the clinical presentation, characteristic histopathological features can confirm the diagnosis. When features are overlapping, immunohistochemistry can be necessary.
Conclusion: since different subtypes of xanthoma have specific clinical and histopathological features and are associated with typical underlying disease, in our opinion histopathologic confirmation, especially in the pediatric patient, is important. This enables accurate referral to a specialized clinician to identify and eventually treat the underlying metabolic or hematologic disease.

Introduction

Xanthomas are benign granulomatous lesions consisting of lipid-laden macrophages. They can be idiopathic or a sign of abnormal lipoprotein metabolism or hematologic disease. They are mostly seen in patients with lipid metabolism disorders or hyperlipidemia which can be primary or secondary [1,2]. The pathogenic mechanism that leads to cutaneous xanthomas is not completely understood. The hypothesis is that when serum levels of lipoproteins are elevated, extravasation of lipoproteins through dermal capillary bloodvessel occurs, followed by digestion by macrophages. This explains the lipid-laden 'foamy cells' that can be found in xanthomas. The process is followed by an inflammatory reaction with the presence of giant cells and fibrosis [3–5]. Some xanthomas occur in patients without signs of dyslipidemia. In these normolipemic patients an association with monoclonal gammopathy, multiple myeloma and other hematologic diseases has been suggested.

Literature distinguishes different clinical variants of cutaneous xanthomas. These different subtypes show different underlying abnormalities, different sites of involvement and different numbers of foam cells or associated histopathologic findings.

Besides xanthelasma, the most common subtype belonging to the subgroup of 'plane xanthomas' with periorbital expression, xanthomas in the head and neck area are very rare [6]. In this case report we describe the case of a young boy with a cutaneous xanthoma in the external acoustic meatus of the right ear. We will resume the current literature on xanthomas in the head and neck area.

Case report

A 11-months-old boy with no significant medical or family history was referred to the out-patient clinic of our tertiary hospital in March 2019 because of a painless, obstructing swelling of the right ear canal since two months. The swelling was itching, bleeding, slightly growing and its color evolved from skin color in the beginning to yellow/red at presentation (Fig. 1). There were no signs of pain, hearing loss or otorrhea. The occurrence of the swelling was not preceded by any trauma. Physical examination revealed a yellowish, subcutaneous swelling of three by 4 mm in the bottom of the right ear canal. There was no opening into the external acoustic meatus cavity. The remaining part of the ear canal skin was normal, the ear drum was intact and the middle ear cavity was air-filled.

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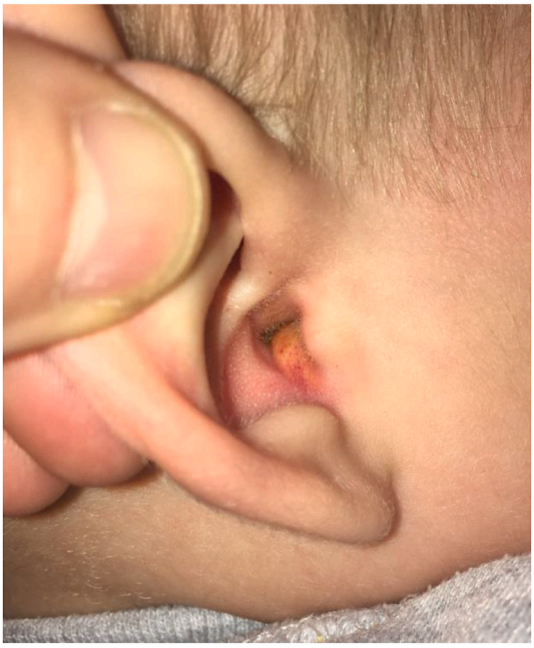

Fig. 1. Image of the obstructive yellowish swelling in the external acoustic meatus at presentation to the out-patient clinic.

Because of the obstruction of the ear canal and for diagnostic purposes we decided to operatively remove the swelling by an endaural approach.

Perioperatively, the swelling consisted of a solid structure with involvement of the skin and was not cystic as was thought in the outpatient assessment. It was attached to the bottom of the ear canal and showed a broad base of attachment to the anterior wall. Because of the benign aspect, radical excision was not pursued. The meatus was covered with an antibiotic gauze dressing for 2 weeks and the postoperative course was uncomplicated with total normalization of the skin after 6 weeks.

Histologic examination revealed non-atypical squamous epithelium with sporadic elongation of the rete ridges. The dermis beneath the epithelium was completely filled with non-atypical histiocytes with regular foamy cytoplasm. These cells are localized between the adnexal structures of the skin and blood vessels.

Sporadically, eosinophilic granulocytes were observed. There were no multinucleated giant cells. Additional immunohistochemistry was positive for CD68 in the lesional cells conforming the presence of histiocytes and macrophages and negative for CD1a en S100, confirming the absence of Langerhans cells. Conclusively, these findings were compatible with xanthoma.

Discussion

The name xanthoma, meaning 'yellow tumor', was first described in 1869 by Smith [7]. These tumors are typically associated with disorders of lipid metabolism. Abnormally high lipoprotein plasma concentration causes permeation of these lipoproteins through the dermal capillary walls. The leaked lipids are then taken up and digested by dermal macrophages evolving to 'foam cells' [5,8]. In primary (heritable) hyperlipidemia, the disturbed lipid metabolism is due to genetic defects in involved receptors, receptor ligands and/or enzymes. In secondary cases, underlying disease or medication is the cause of hyperlipemia. Examples of diseases include obesity, diabetes mellitus, hypothyroidism, alcohol abuse, biliary tract obstruction and nephrotic syndrome. Medication leading to hyperlipidemia includes estrogens, prednisone and cyclosporine among others [6]. Seldom, xanthoma occur in normolipemic patients explained by different hypotheses including altered lipoprotein content or structure, divergent lipid storage or underlying lymphoproliferative disease with xanthomatization (lipid accumulation) of cells infiltrating the dermis [6,8].

Cutaneous xanthomas are most often discovered in adulthood and males and females are equally affected. In patients with familial hypercholesterolemia xanthomas can develop prior to the age of 10. Accurate diagnosis is of great importance to identify and eventually treat underlying disease.

Xanthomas mostly present as erythematous to yellow papules, plaques or nodules. The amount of lipid present and its depth below the surface determines the color of the lesion. The characteristic histologic feature is the presence of lipid-laden macrophages also known as 'foam cells'. The number of foam cells and presence of associated histopathological features such as inflammatory cells, extracellular lipid deposition and fibrosis varies between the different subgroups and the age of the xanthomas [8].

The different clinical variants include the eruptive (mostly extensor surfaces of the extremities and buttocks), tuberous (mostly over joints or extensor surfaces especially elbows and knees), tendinous (over tendons or ligaments, mostly the Achilles tendon), plane (neck, trunk, shoulders and axillae and around eyelids as xanthelasma), verruciform (oral cavity or anogenital skin) and papular (face, trunk and mucous membranes) xanthomas, the last being very rare. Only the verruciform type is not associated with hyper- or dyslipidemia or underlying disease but is thought to be caused by an immune reaction to local trauma or inflammation [6]. Additionally, in literature, few cases are described with the presence of isolated xanthomas which cannot be fitted into the above mentioned classification. Furthermore, the group of xanthogranulomas is specifically distinguished from the xanthomas since they are not related to lipoprotein metabolism disorders and consist of a mixture of cell types [8].

Assessing for risk factors (underlying disease, medication, family history) is recommended in all patients. The presumable diagnosis can be made by assessing the classic clinical features on physical examination and potentially confirmed by histopathologic findings.

Except for those patients with obvious verruciform xanthomas, in all patients a fasting lipid panel to assess for dyslipidemia is advised. In normolipidemic patients with plane xanthomas screening for associated hematologic disease is recommended [6].

Most patients can be cured without surgery by identifying and medically treating the underlying cause. In patients with hyperlipidemia, the focus of treatment is a change in diet with restriction of saturated fats and cholesterol. Pharmacotherapy is indicated when diet alterations are not sufficient [5,2]. In normolipidemic patients surgical or destructive therapies such as cryotherapy are indicated in case of cosmetic complaints. Furthermore, surgical removal is indicated for diagnostic purposes or when there are local symptoms. Complete excision is then advised to prevent continued growth of remaining tissue [7].

Literature on xanthoma involving the temporal bone is rare and even lacking in the pediatric population. The only existing literature review reports of only eleven cases with an age ranging from 33 to 67 years. Xanthoma in the external ear canal was reported in one case. In all patients, the serum cholesterol concentration was increased and dietary alternations was the main focus of treatment. It was furthermore shown that xanthoma may reach gigantic proportions with minimal clinical symptoms. In one case describing an expanded xanthoma of the petrous apex, the extent of bone destruction was such that vital structures (carotid artery, jugular vein and multiple cranial nerves) were surrounded without disturbing their function. The lesion in that case represented the first and only clinical manifestation of the patient's hyperlipidemia [1].

The differential diagnosis of unilateral ear canal masses includes juvenile xanthogranuloma, leiomyoma, hemangioma, neurofibroma, osteoma, lipoma and fibroma. These benign soft tissue tumors are mostly easy to distinguish based on their clinical features and diagnosis can be

confirmed by histopathologic examination. In some cases, immunohistochemistry is necessary to conform the diagnosis when features are overlapping [9,10].

Without proper treatment, xanthomas tend to persist. Follow-up is necessary for patients in case of hyperlipidemia to prevent for further morbidity and for patients with plane xanthomas with no signs of hematologic disease at the time of diagnosis, to recognize eventual development of hematologic disease.

In our patient, thorough and revised histopathologic examination and immunohistochemistry confirmed the uncommon presence of a xanthoma. The clinical features (yellowish/red swelling) and localization of the lesion fits best with the group of planar xanthomas. The absence of typical giant cells, neutrophils and lymphocytes makes the presence of a juvenile xanthogranuloma less likely. He was referred to the pediatrician for further evaluation of his lipid profile and laboratory testing for the presence of hematologic disease. This showed no abnormalities up to this date, nor were there any signs of local abnormalities seen in the externa ear canal during the last visit to our outpatient clinic in February 2020.

Patient consent

Consent to publish the case report was not obtained. This report does not contain any personal information that could lead to the identification of the patient.

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Authorship

All authors attest that they meet the current ICMJE criteria for Authorship.

Declaration of competing interest

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

References

[1] Jackler RK, Brackmann DE Xanthoma of the temporal bone and skull base. Am J Otol 1987;8:2.
[2] Shinnabe A, Tamii S, Hara M, et al. Xanthoma in the middle ear cavity. Otology & Neurology 2013;34:53–4.
[3] Walton KW, Thomas C, Dunkerley J The pathogenesis of xanthoma. J Pathol 1973; 109:271–89.
[4] Friedman O, Hockstein N, Willcox Jr TO, et al. Xanthoma of the temporal bone: a unique case of this rare condition. Ear Nose Throat J 2000;79:433–6.
[5] Pitman M, Elhosseiny A, Moscattelo AL Xanthoma of the maxillary sinus. Otolaryngol Head Neck Surg 2002;126:426–7.
[6] Wanat K, Noe M. Cutaneous xanthomas. In: UpToDate (last updated October 21, 2019).
[7] Rosenberger HC Solitary xanthoma of the external auditory canal. Arch Otolaryngol 1937;26:395–9.
[8] Weedon D, Strutton G, Rubin AI, Weedon D Weedon's skin pathology 2010;961–5.
[9] Hock M, Zelger B, Schweigmann G, et al. The various clinical spectra of juvenile xanthogranuloma: imaging for two case reports and review of the literature. BMC Pediatr 2019;1:128.
[10] Gaffar BA, Manzigi M, Saleem N Benign mesenchymal tumors of the external ear: a series of 14 cases. Ann Diagn Pathol 2019;41:62–8.