Received: December 28, 2022
Accepted: February 27, 2023

Acad Med J 2023;3(1):147-151
UDC: 616.5-006.2-056.7-055.1-053.6
www.doi.org/10.53582/AMJ2331147s
Case report

STEATOCYSTOMA MULTIPLEX IN AN ADOLESCENT BOY: A CASE REPORT

Sotirovski Tomica[1], Tusheva Ivana [1], Blazeska Ana[1], Dzambazova Marija[1], Peev Igor[2], Damevska Katerina[1]

[1]University Clinic for Dermatology, Faculty of Medicine, Ss. Cyril and Methodius University, Skopje, Republic of North Macedonia
[2]University Clinic for Plastic and Reconstructive Surgery, Faculty of Medicine, Ss. Cyril and Methodius University, Skopje, Republic of North Macedonia
e-mail: tomicasot@gmail.com

Abstract

Steatocystoma multiplex (SM) is a rare genodermatosis transmitted as an autosomal dominant trait caused most often by a mutation in the gene coding for keratin 17. There are some sporadic cases described in the literature. The disease commonly manifests as numerous intradermal cysts caused by hamartomatous malformations of the pilosebaceous duct junction. The cysts can be located in any skin region, mainly on the chest. The disease is usually benign and asymptomatic, but it can be exceptionally disfiguring, which is the main reason for a medical visit. Pachyonychia congenita type-2 and the eruptive vellus hair cyst are closely related to SM; therefore, histopathological confirmation is necessary before starting any treatment. Here, we present a case of a widespread SM in an adolescent boy, focusing on its clinical and histopathological characteristics.

Keywords: steatocystoma multiplex, keratin-17, hereditary, cyst, acne, genodermatosis

Introduction

Steatocystoma multiplex (SM) is a rare genodermatosis transmitted as an autosomal dominant trait caused most often by a mutation in the gene coding for keratin 17 (KRT17). This keratin protein is found in the nail bed, hair follicles, and sebaceous glands. Mutations in KRT17 can also result in one form of paronychia congenita, in which cysts, plantar keratoderma, and natal teeth are common[1]. The defective keratin network disrupts the growth and function of cells in the skin and nails, including cells that make up the sebaceous glands. These abnormalities lead to the development of sebum-containing cysts. There are some sporadic cases described in the literature. SM is hamartomatous manifestation of the pilosebaceus duct junction that commonly manifests as numerous intradermal cysts that develop in puberty[2]. The disease is divided into generalized, localized, facial, acral, and suppurative types. In males, the disease most often manifests on the chest. The lesions are usually 2-20 mm in diameter, dome-shaped, translucent, or white to yellowish. Sometimes a small central punctum can be identified, containing one or more hairs. Spontaneous rupture will result in steatocystoma multiplex suppurativum, which might be mistaken for acne conglobate[1,2].

SM is a benign disease. However, the quality of life may be diminished as some patients may have psychological implications resulting from the disfigurement due to widespread lesions or scarring in the inflammatory variant. No reports describe malignant transformation of SM[3].

Case report

A 16-year-old boy of Roma ethnicity visited the clinic because of numerous nodular and cystic lesions on the chest that had been present for a few years. Previously, these lesions were diagnosed as nodulocystic acne. Various treatments have been tried, including several courses of systemic antibiotics, but none showed a particular effect. The patient was adopted, and therefore the family history was unknown.

On examination, the dermal cysts were well defined, round to oval, whitish colored, with smooth surface, and different in size (2-5mm) (Figure 1 and 2). Besides the case being clinically apparent, an excisional biopsy was advised to confirm the diagnosis. Histopathology revealed a cyst wall lined by stratified squamous epithelium without a granular layer with a corrugated eosinophilic cuticle surface. Multiple sebaceous glands were identified adjacent to the cyst wall, pathognomonic for steatocystoma.

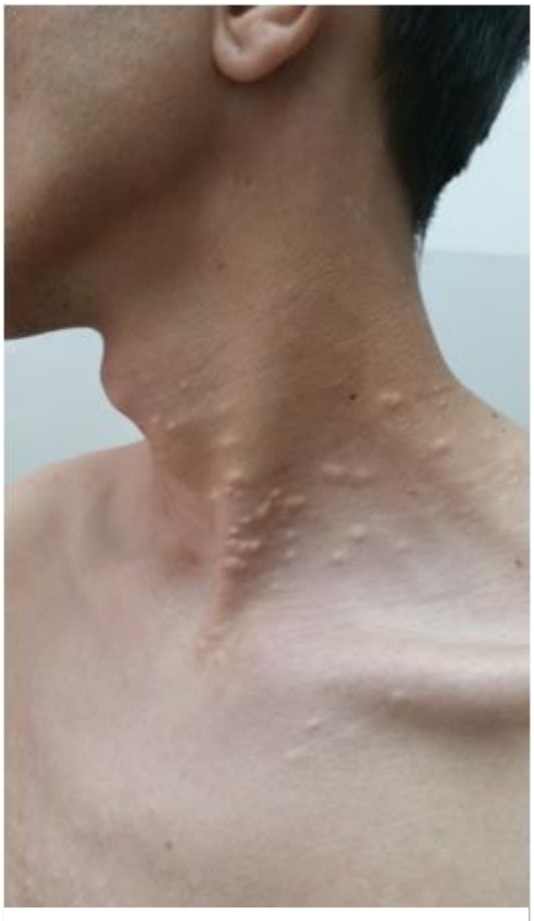

Fig. 1. Whitish, round to oval cystic lesions on the neck with 2-5 mm in diameter. First examination

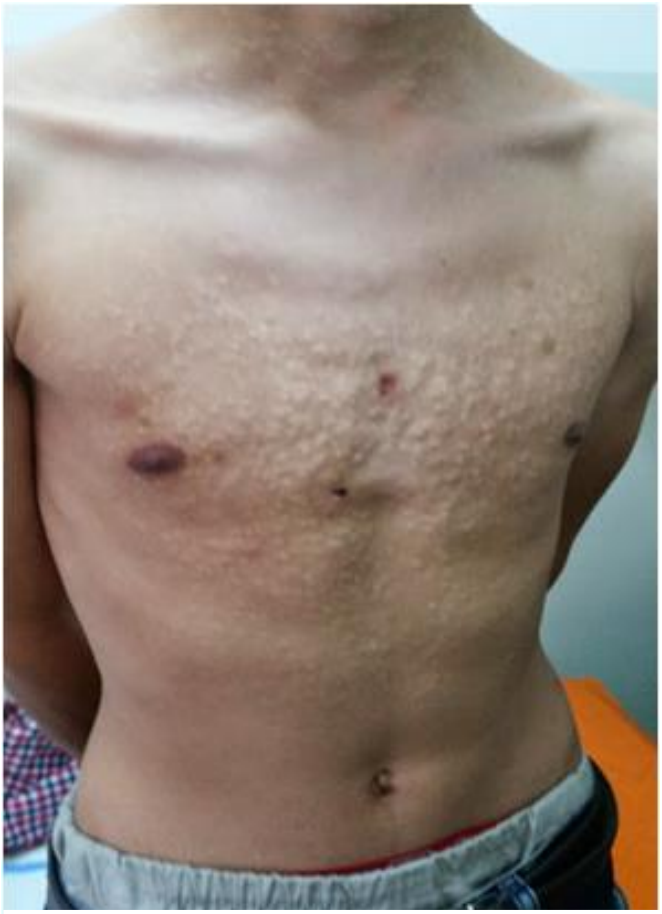

Fig. 2. Multiple translucent flesh-colored to yellow dome-shaped papules and nodules on the chests. First examination.

His disease steadily advanced; at age 18, he had more than 200 cysts involving the neck and chest, while his face and acral regions remained spared. The lesions were asymptomatic but cosmetically disfiguring , without a tendency for exulceration. At the age of 19, a therapeutic attempt was made on selected neck lesions; several lesions were excised under local anesthesia, and some cysts were punctured with the evacuation of the contents. Because of the number of cysts, the excision technique was difficult and unpleasant for our patient.

Discussion

Steatocystoma multiplex (SM; OMIM184500) is a rare genetic skin disease that results from a mutation of the keratin 17 gene (KRT17; OMIM148069) on chromosome 17q21.2.1[3,4]. This same genetic mutation also causes pachyonychia congenita type 2 (PC-2). Patients with PC-2 have milder keratoderma, natal teeth, pili torti, angular cheilosis, and hoarseness. PC-2 patients have multiple cysts, some of which are steatocystomas[4]. In addition, persistent infantile milia may sometimes be associated with both SM and eruptive vellus hair cysts and may represent a disorder with a predisposition to multiple pilosebaceous cystic lesions.

Pringle coined the term steatocystoma multiplex in 1899 after being described by Jamieson in 1873[1].

Clinically, the disease is characterized by the progressive appearance of skin-colored subcutaneous cysts. It may occur at any site, but the trunk is the most common location. The cysts are located principally on the upper anterior portion of the trunk, upper arms, axillae, and thighs. The sternal region is commonly affected in males, and axillae and groin are widely affected in female patients. The lesions lack a punctum. The solitary lesions are sporadic, known as Steatocystoma simplex[2].

According to the site involvement, the disease is divided into generalized, localized, facial, acral, and suppurative types[4].

Lesions usually appear in adolescence or early adulthood, when sebaceous activity is at its peak. However, the development of SM can first occur in late adulthood also. In severe cases, the lesions may be generalized, sparing only the palms and soles. At times the lesions may be limited to the face or scalp, a distinct form termed a papular facial variant[4,5]. Lesions limited to the genital area have also been reported[1,4]. Congenital and adolescent-onset linear lesions are rare. Steatocystoma may be larger (up to 2 cm) and prone to rupture and suppuration (steatocystoma multiplex suppurativum). If these lesions are widespread, the condition can be very disfiguring. Steatocystomas contain a syrup-like, yellowish, odorless, oily material. In the suppurative type, colonization with bacteria can occur, leading to foul odor and social isolation[4,6].

Steatocystoma multiplex consists of benign cystic lesions derived from the sebaceous glands. It is well known that in the etiopathogenesis of the disease, multiple hamartomatous malformations of the pilosebaceous duct junction (hair follicle unit) develop at puberty. Hormones stimulate sebaceous glands to increase sebum production, which might also be the reason for pubertal onset[7]. In line with this fact and along with the macroscopic look of the lesions, the diagnosis can be clinically confused with acne conglobata or milia besides the different etiology. Histopathological examination is the key to a precise diagnosis[8].

Cysts in exposed areas of the body create serious self-image problems. Treatment options for SM are somewhat limited. Individual cysts can be removed surgically, either by excision or by incision and draining its contents[9,10]. Because of the number of cysts and their fragility, standard cyst excision techniques are impractical. Pamoukian and Westreich[10] described a

modified excision method that yielded good results. Their approach consisted of stab incision into the cyst and manually evacuating its contents. Afterwards, a fine hemostat was inserted to grasp the cyst wall from within and strip out its lining. In one sitting, 50 to 150 cysts could be removed.

Lasers, electrosurgery, or cryotherapy can also be used. Inflammation can be reduced with oral antibiotics [7,11-13]. Oral isotretinoin is not curative but may help shrink the cysts and reduce inflammation[10].

In conclusion, we present a rare case of a boy with SM that was previously diagnosed and treated as acne vulgaris. Definitive diagnosis was made with excisional biopsy and histopathology. The surgical approach was very uncomfortable for the patient and deemed unacceptable. That is why we suggest laser treatments as a better option in the future. Reporting rare cases such as SM will help us understand and determine the incidence of the disease.

Conflict of interest statement. None declared.

References

1. Shin NY, Kang JH, Kim JE, Symkhampa K, Huh KH, Yi WJ, *et al*. Steatocystoma multiplex: A case report of a rare entity. *Imaging Sci Dent* 2019; 49(4): 317-321. doi: 10.5624/isd.2019.49.4.317.
2. Amin M, Hashim P. Steatocystoma Multiplex: Case Report and Review of Treatment. *SKIN The Journal of Cutaneous Medicine* 2018; 2(1): 75-79. doi.org/10.25251/skin.2.1.12.
3. Georgakopoulos JR, Ighani A, Yeung J. Numerous asymptomatic dermal cysts: diagnosis and treatment of steatocystoma multiplex. *Can Fam Physician* 2018; 64(12): 892-899. PMID: 30541803; PMCID: PMC6371868.
4. Kamra HT, Gadgil PA, Ovhal AG, Narkhede RR. Steatocystoma multiplex-a rare genetic disorder: a case report and review of the literature. *J Clin Diagn Res* 2013; 7(1): 166-168. doi: 10.7860/JCDR/2012/4691.2698.
5. Chu HD. Steatocystoma multiplex. *Dermatology Online J*. 2013; 9(4):18. 2013 doi: 10.7860/JCDR/2012/4691.2698.
6. Wang X, Shi Y, Ye Y, Liu F, Jin W, Chen W, *et al*. Keratin 17 gene mutation in patients with steatocystoma multiplex. *Zhonghua Yi Xue Za Zhi* 2001; 81(9): 540-543. PMID: 11809119.
7. Choudhary S, Koley S, Salodkar A. A modified surgical technique for steatocystoma multiplex. *J Cutan Aesthet Surg* 2010; 3(1): 25-28. doi: 10.4103/0974-2077.63284.
8. Kamra HT, Gadgil PA, Ovhal AG, Narkhede RR. Steatocystoma multiplex-a rare genetic disorder: a case report and review of the literature. *J Clin Diagn Res* 2013; 7(1): 166-168. doi: 10.7860/JCDR/2012/4691.2698.
9. Gass JK, Wilson NJ, Smith FJ, Lane EB, McLean WH, Rytina E, *et al*. Steatocystoma multiplex, oligodontia and partial persistent primary dentition associated with a novel keratin 17 mutation. *Br J Dermatol* 2009; 161(6): 1396-1398. doi: 10.1111/j.1365-2133.2009.09383.x.
10. Antal AS, Kulichova D, Redler S, Betz RC, Ruzicka T. Steatocystoma multiplex: keratin 17 - the key player? *Br J Dermatol* 2012; 167(6): 1395-1397. doi: 10.1111/j.1365-2133.2012.11073.x.

11. Pamoukian VN, Westreich M. Five generations with steatocystoma multiplex congenita: a treatment regimen. *Plast Reconstr Surg* 1997; 99(4): 1142-1146. doi: 10.1097/00006534-199704000-00036.
12. Kim SJ, Park HJ, Oh ST, Lee JY, Cho BK. A case of steatocystoma multiplex limited to the scalp. *Ann Dermatol* 2009; 21(1): 106-109. doi: 10.5021/ad.2009.21.1.106.
13. Covello SP, Smith FJ, Sillevis Smitt JH, Paller AS, Munro CS, Jonkman MF, *et al*. Keratin 17 mutations cause either steatocystoma multiplex or pachyonychia congenita type 2. *Br J Dermatol* 1998; 139(3): 475-480. doi: 10.1046/j.1365-2133.1998.02413.x.