ABSTRACT

Objective: Pilomatrixoma is a benign skin tumor. The aim of this study is to describe the clinical presentation and associated conditions in children with pilomatrixoma.

Methods: The medical records of 52 children from a single referral center obtained between 2000 and 2016 were retrospectively reviewed.

Results: There were a total of 62 tumors in 52 children. The mean age at excision was 9.55±4.65 years. Tumors were predominantly located in head and neck region (48.4%). There was no family history of pilomatrixoma, except one case. One patient had Turner Syndrome and the other one had tuberous sclerosis complex. Fifty-four (87%) lesions were examined by ultrasonography (USG). Pilomatrixoma was considered in the differential diagnosis in eight patients (15.3%) by a radiologist.

Conclusion: Pilomatrixoma is one of childhood benign skin tumors which could be detected by superficial USG method in children. It should be kept in mind for differential diagnosis in children with superficial masses.

INTRODUCTION

Pilomatrixoma is a benign skin tumor which was first described by Malherbe and Chenantais in 1880. It was called “calcifying epitheliomas of Malherbe” because it was thought to have originated from sebaceous glands at that time. In 1961, Forbis and Helwig found that the outer root sheath cell of hairy follicle was actually its source of origin. Since 1977, the terms of “pilomatrixoma” or “pilomatricoma” have been globally accepted to name the lesion. Pilomatrixoma is a benign tumor typically presents in childhood, particularly within the first decade of life. The adult-onset type of pilomatrixoma has been also defined but it is often associated with nonspecific malignancies. Thus, a bimodal pattern of occurrence has been reported with the first peak seen at 5-15 years and the second peak seen at 50-65 years. The head and neck are the most common body regions for a pilomatrixoma. Cases outside of the head and neck region are commonly associated with genetic syndromes and disorders. The only curative treatment of pilomatrixoma is complete excision.
The aim of this study is to (i) present our institutional experience with pilomatrixoma, describing its clinical presentation, associated conditions, radiologic and pathologic findings, and (ii) attract attention to this tumor for differential diagnosis of benign skin masses in children.

MATERIAL and METHODS

The medical records of 52 children with histologically diagnosed pilomatrixoma at plastic and reconstructive surgery department of our institution between 2000 and 2016 were evaluated retrospectively. The electronic medical records of the patients consisted of the patients’ clinical medical history and pathological specimen results. Ethical approval for this study was obtained from Mersin University Faculty of Medicine (IRB No. 2000-89-102). The demographics included the patients’ sex, age at operation, location of the mass, number of mass lesions (solitary or multiple), radiological findings, complications, and recurrence of the lesions. Radiological imaging results were also reviewed to analyze the characteristics of pilomatrixoma and determine their diagnostic accuracy.

Statistical Analysis

Statistical analyses were performed using the SPSS software version 24 (IBM Corp. Released 2016. IBM SPSS Statistics for Windows, Version 24.0. Armonk, NY: IBM Corp.). Kolmogorov-Smirnov test was used to determine the normal distribution of numerical variable. Categorical data were presented with numbers (n) and percentages (%), and numerical data with mean±standard deviation (SD) and minimum-maximum (min-max). The Pearson correlation test was used for investigating a correlation between numerical data. Type I error was determined as 5% and a p value of <0.05 was considered statistically significant.

RESULTS

Medical charts of 52 pediatric pilomatrixoma patients [24 (46.2%) male and 28 (53.8%) female] were reviewed retrospectively. The mean age at excision was 9.55±4.65 years (1-17 years). The mean tumor diameter was 2.17±1.24 cm (0.5-5.5). One of the patients (1.9%) had a family history of pilomatrixoma. One patient (1.9%) had Turner syndrome and one (1.9%) had tuberous sclerosis complex.

Multifocal disease was detected in 4 children (7.7%). The median number of synchronous lesions was 4 (3-7). Two patients (3.8%) had metachronous pilomatrixomas. The localization of 62 lesions were as follows; head and neck (n=30, 48.4%), upper limbs (n=15, 24.2%), trunk (n=11, 17.7%) and lower limbs (n=6, 9.7%). Cervical (n=14, 22.6%), preauricular (n=6, 9.7%), scalp (n=4, 6.5%) lesions, and one lesion (n=1, 3.3%) on the chin, parotid region, upper eyelid, lower eyelid, nose and cheek were surgically excised. One (1.6%) lesion was located on the areola of the breast.

The most common clinical presentation was asymptomatic, slowly growing, subcutaneous mass attached to the skin (n=56, 90.3%). The mass was mostly hard in tenderness but it was freely mobile when stretched (n=56, 90.3%). However, two children (3.8%) had discomfort and one (1.9%) had rapidly growing mass. Three patients (5.8%) had complication of acute infection. The median duration of the lesions was 6 months (6 weeks-3 years).

Fifty-four (87%) lesions were examined by ultrasonography (USG). All tumors (100%) were located in the subcutaneous layer. The median diameter of tumor was 12.8 mm (5.6-59) measured by USG. Forty-two (77.8%) lesions had oval and 12 (22.2%) lesions had irregular shape. Forty-six (85.2%) lesions were hypoechoic and eight (14.8%) lesions were hyperechoic. Most of the lesions were also heterogeneous. All lesions had posterior shadowing. Peripheral hyperechoic rim was observed in 48 (88.9%) lesions. Echogenicity, echo texture, margin, and hyperechoic rim could not be evaluated in six (9.7%) cases of pilomatrixoma. Doppler flow signals were observed in the peripheral region in 38 (70.3%) lesions and in two (3.2%) lesions in the central region. There was no correlation between mass size and the region where Doppler flow signals were obtained (p>0.05). Pilomatrixoma was considered in the differential diagnosis of eight patients (15.4%) by radiologist.

The diagnosis of pathological specimens were confirmed as pilomatrixoma by pediatric pathologist in all cases except one which was diagnosed as...
dermoid cyst (Figure 1A and 1B). Epidermal cyst like structures undergoing keratinization with central keratin debris and foreign body giant cell inflammatory reaction to keratin debris were present in some cases (Figure 1C) and the calcification was seen in most cases (Figure 1D).

Recurrence of the disease was observed in two (3.8%) subjects and no recurrence was detected during the 4-year follow-up after resection.

**DISCUSSION**

In this observational study, the majority of tumors occurred before 10 years of age. The prevalence of pilomatrixoma is common in females, with a female-to-male ratio of 1.5-2.06.

Pilomatrixoma is typically present as a superficial, slowly and growing painless mass. It is firm but freely mobile in texture, mostly lobulated in nature, and fixed to the epidermis. The overlying skin is usually normal in appearance. Similar to our findings, they occur mostly in the head and neck region, but rarely they can be found at any hair-bearing site, upper extremities, trunk, and lower extremities. In parallel with the literature, we observed female-male ratio around 1.2 to 1.0.

Multiple pilomatrixomas are rarely seen, but they are mostly associated with genetic disorders including familial adenomatous polyposis, myotonic muscular dystrophy, Rubinstein-Taybi syndrome, Turner syndrome, Kabuki syndrome, and childhood cancer syndrome constitutional mismatch repair deficiency.
Neurofibromatosis type 1 was also diagnosed in patients with sporadic pilomatrixoma. To the best of our knowledge, tuberous sclerosis complex was not reported to date. In this context, we reported the first case with tuberous sclerosis complex accompanying pilomatrixoma. According to Schwarz et al. the incidence of multiple lesions was 8.2-33.3%, and the lesions may be synchronous or metachronous. The incidence of multiple pilomatrixoma was 7.69% and only one patient had a family history without any underlying disease in this series.

Bulman et al. reported the diagnostic accuracy of pilomatrixoma by USG as 13.3%. In present study, only eight patients (15.3%) were correctly diagnosed by USG. We suggest that USG may provide limited benefits in the differential diagnosis of such lesions. It is reported that hypoechoic lesions are the most common features, as in our study. In previous studies, the prevalence of hypoechoic rim that represents the capsule of the pilomatrixoma, was found between 65% and 75% of the cases whereas in our study it was determined as 88.8%. In this study, Doppler USG examination could be performed in 64.5% of the lesions and vascularity was observed in 70.3% of these lesions. This is similar to the reported vascularity of 50-70% in pilomatrixomas in pediatric population.

Pathological diagnosis of pilomatrixoma could be done with fine-needle aspiration biopsy (FNAB) or with total surgical excision. Although histological findings of pilomatrixomas are well-known, FNAB is accepted as an important method for preoperative diagnostic research. However, the cytologic diagnosis of pilomatrixoma is sometimes difficult and they are misdiagnosed. For this reason, we preferred biopsy to make the diagnosis more accurately. Pathologically, there are two basic cell types, basophilic cells and eosinophilic shadow cells with an intervening connective tissue stroma containing blood vessels, foreign-body giant cells, mixed inflammatory cell infiltration and sometimes hemosiderin and rarely amyloid. We observed eosinophilic shadow cells toward the central areas of the cell masses predominantly. Calcification occurs in more than two-thirds of the tumors and is usually in the shadow cells. Calcification of the stroma occurs in about 13%; hemosiderin is found in about 25% of cases; and melanin is present in nearly 20% of lesions and may be in the shadow cells as well as in the stroma. No calcification of the stroma was observed in our population.

Although pilomatrixoma is one of the most common cutaneous tumors in children and adolescent, it is usually not considered in the differential diagnosis of pediatric head and neck masses. The rate of preoperative diagnostic accuracy of pilomatrixomas ranges from 0 to 49 percent. Similarly, in our most of the patients, the pilomatrixoma was not considered in differential diagnosis, only eight patients (15.3%) had correct preoperative diagnosis by USG.

In conclusion, we suggest that pilomatrixoma should be considered in the differential diagnosis of superficial or subcutaneous masses which particularly located at head and neck, particularly in children. Surgical excision of the pilomatrixoma is recommended for definitive diagnosis and curative treatment.

Acknowledgements: None

Ethics Committee Approval: For this study, ethical approval was obtained in 2019 Mersin University Faculty of Medicine (IRB No. 2000-89-102).

Conflict of Interest: The authors declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

Funding: The authors received no financial support for the research, authorship, and/or publication of this article.

Informed Consent: Parents of the patient provided informed consent to publish the report.

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