ACTA
ORTHOPAEDICA
et
TRAUMATOLOGICA
TURCICA

Author's translation
Acta Orthop Traumatol Turc 2005;39(4):365-368

Familial glomus tumor encountered in the same finger and localization in four family members

Ailenin dört üyesinde aynı parmak ve yerleşimde görülen glomus tümörü

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Sağ el üçüncü parmağında aşırı duyarlılık ve şiddetli ağrı yakınması ile birçok merkeze başvurduktan sonra bir beyin cerrahı tarafından kliniğimize sevk edilen 35 yaşındaki erkek hastanın öyküsünden yakınmalarının üç yıldır devam ettiği öğrenildi. Sağ el orta parmağının eponişyumu altında hassas, çok ağrılı bir kitle palpe edildi. Kitle glomus tümörü öntanısıyla cerrahi olarak çıkartıldı ve tanı histolojik incelemede doğrulandı. Hastanın aile öyküsünden, aynı şikayetlerin babası ve iki kız kardeşinde de olduğu ve yine sağ el üçüncü parmağın tutulduğu öğrenildi. On yıl önce ameliyat olan babanın kayıtlarının incelenmesi ve kız kardeşlerin muayenesindeki klinik-radyolojik bulgular aynı tanıyı destekliyordu. Parmakta ağrı ve hassasiyet gibi yakınmalar olduğunda, aile bireyleri de sorgulanmalı ve ayırıcı tanıda ailesel glomus tümörü akla getirilmelidir.

Anahtar sözcükler: Parmak/cerrahi; glomus tümörü/genetik/patoloji/cerrahi; el/cerrahi; deri neoplazileri/cerrahi; yumuşak doku neoplazileri/cerrahi.

A 35-year-old man was referred to our clinic by a neurosurgeon for hypersensitivity and severe pain in his right middle finger. History showed that the symptoms had been present for three years and he had made several attempts to seek medical attention. A sensitive, very painful mass was palpated under the eponychium of his right middle finger. The tumor was surgically removed and histological examination confirmed the clinical diagnosis of a glomus tumor. Inquiry into the family history revealed that the same problem existed in the same localization with similar complaints in the father and two daughters. Both the records of the father's operation 10 years before and clinical-radiological findings of the daughters were consistent with a familial entity. In the presence of hypersensitivity and pain in the finger, family members should be inquired and a diagnosis of familial glomus tumor should be considered.

Key words: Fingers/surgery; glomus tumor/genetics/pathology/surgery; hand/surgery; skin neoplasms/surgery; soft tissue neoplasms/surgery.

Glomus tumor arises from glomus body which is an arteriovenous anastomosis and controls the blood flow of skin. It is also known as glomuvenous malformation, "glomus cell venous malformation" or "glomangioma". Although being a benign tumor with unknown etiology and generally occuring sporadically; glomus tumor has been reported to be inherited in an autosomal dominant genetic pattern.[1-3] Differently from sporadic type, inherited glomus tumor with slowly progressing clinical picture eventually becomes a hypersensitive mass.[4] It is one of the rarely encountered characteristics of the familial glomus tumor to be found in the same finger of family members and even in the same localization.

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Received: 22.06.2004 **Accepted:** 26.02.2005

Classic symptoms of a glomus tumor are pain, tenderness and hypersensitivity to temperature changes (clinical triad).[5] Glomus tumors are usually located in the distal segment of the fingers under the nail matrix or in the pulpa. Symptoms may have been persisting for a long time because of failure in diagnosis and treatment. Changes in temperature, palpation, and touching may cause pain and hypersensitivity.[6] When located in deep layers like in pulpa, it is difficult to make a diagnosis because of less obvious hypersensitivity there is no palpable mass and the extent of hypersensitive area is almost a pencil point wide. These patients without objective findings usually do not accept an operation.[7] Transillumination[8], MRI[9] and US[6] are useful tools to confirm the tumor location. As long as there is no bony erosion, plain X-Rays can not be helpful.[10] To provide an immediate relief of the symptoms, the most effective treatment method is surgical excision. The incidence of recurrence after surgical excision ranges from 5 to 50%.[11] Early recurrence, therefore, is usually attributed to incomplete excision,[5] while late recurrence to be a new tumor formation. [11]

Case report

A 35 year old man, referred to our clinic by a neurosurgeon, had been suffering from hypersensitivity and severe pain of his right middle finger for the last three years. In the physical examination, there was a little, sensitive palpable mass under eponychium. When palpated, the pain was so severe that the patient was fainting. The pulpa and nail were normal in appearance. Plain X-Rays showed no spesific findings.

Initial diagnosis was benign soft tissue tumor and to provide complete excision, surgical exploration was offered to the patient. Under regional intravenous anesthesia, using surgical loop (x4.5), eponichium of third finger of the right hand was elevated through bilateral 1 cm vertical incisions. By lifting nail and nail matrix, the soft tissue tumor in 4 mm diameter was removed (Fig.1). Histological examination confirmed the diagnosis of glomus tumor. The pain and hypersensitivity disappeared immediately after surgery.

When family history was questioned, the father was told to have undergone a few consecutive operations in the same finger because of the similar symptoms ten years ago and the histological diagnosis was glomus tumor. Additionally, two daughters have been suffering from same complaints in their same finger for three and five years respectively. Similarly, they also had had a few failed diagnosis and treatment attempts. They asked to attend our clinic for clinical and radiological examination. The localization of pain was the same site of right middle finger in both of them. The pain and hypersensitivity were very severe on the volar aspect of distal phalanx of right middle finger but there was no palpable mass. There were no findings on X-Ray and MRI to explain the clinical picture. Ultrasonography was performed to reveal the condition that causing the clinical symptoms. An US examination performed with a 6-9 MHz linear transducer (GE Medical Sys, Logiq 500, Pro Series) showed a two-milimeter diameter, round and hypoechoic lesion on the volar aspect of the tip of distal phalanx (Fig. 2).

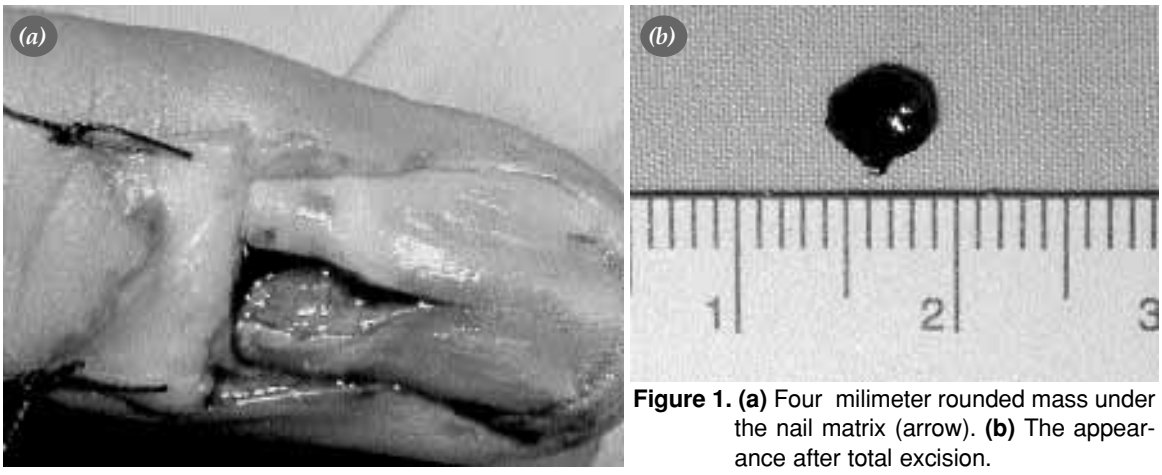

Figure 1. (a) Four milimeter rounded mass under the nail matrix (arrow). **(b)** The appearance after total excision.

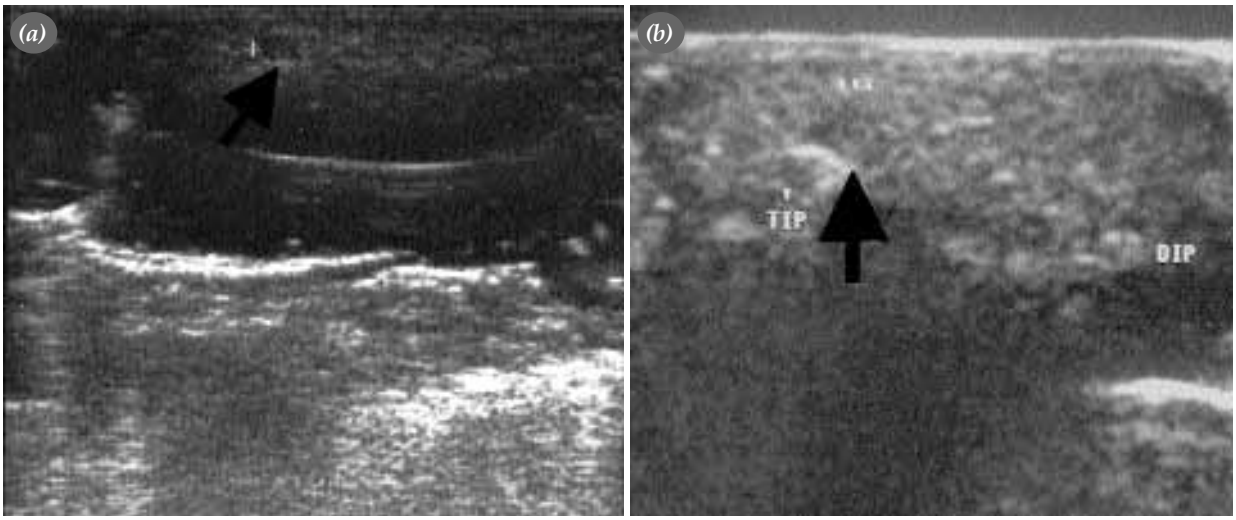

Figure 2. US examination of two daughters respectively. **(a and b)**. Two milimeter diameter, hypoechoic lesion just on the tip of distal phalanx (arrows). TIP: Tip of distal phalanx. DIP: Distal interphalangeal joint.

They were offered surgical exploration, suspecting glomus tumor. Since they had no pain as severe as that of their brother's and had no palpable, obvious mass, they did not accept the operation.

Discussion

Since, usually, glomus tumor is too deep, it is hardly palpable. Therefore in the presence of clinical triad, glomus tumor should be suspected. In addition to clinical triad, familial inheritance and distal phalangeal localization should direct the clinician towards the diagnosis. However definitive diagnosis can only be established by histologic examination. The reason of the high rate of failed diagnosis and treatment attempts is lack of suspicion during examination of the patient with unpalpable glomus tumor. Just as it is difficult to convince the patient with unpalpable mass to take an operation, so too it is difficult to asess the localization of tumor during the surgery of the patient under anesthesia. [5] If there is discoloration on the nail and palpable sensitive mass, it is easy to determine the location of tumor. It is important to know the tumor location and size for avoiding incomplete excision which is the most important factor in recurrence.

The reason of glomuvenous malformations is mutations that took place in glomulin gene which encoded as 1p21-22. [138000 OMIM] The glomulin gene is 19 exon and 55 kb. [12] Glomus tumor is transmitted in an autosomal dominant pattern via the paternal line. [168000 OMIM] Van der Mey et al [13] have thought autosomal dominant gene to be inactivated during oogenesis and thus tumor development was avoided in the next generation. Therefore a daughter whom inherited the disease from father, can not inherit it to her children. Consequently; familial glomus tumor, can be seen in different locations or in the same finger and location in the family members. In the presence of similar clinical findings and paternal transmission, familial glomus tumor should be suspected.

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