

A case of isolated macrodactyly

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Dear Editor,

Macrodactyly can be used synonymously with megalodactyly or digital gigantism. It is characterized by the congenitally large formation of one or more fingers of the hand or foot. Macrodactyly is a rare anomaly. There is a growth in the affected soft tissue and bone. It can clinically cause pain and loss of function, stiffness, finger-tip ulceration, carpal tunnel syndrome in hand (1). Patients diagnosed with macrodactyly should be screened for diseases such as Milroy's disease, Proteus syndrome and Klippel-Trenaunay-Weber syndrome. Diagnosis and treatment planning are performed by imaging methods (1,2,3).

A 23-month-old male infant presented to our outpatient clinic because of congenital growth in his left hand index finger and thumb. Physical examination revealed prominence in the left thumb, swelling of the index finger and medial folding of the left index finger (Figure 1). There was no abnormality in other parts of the body. There was no tooth and nail pathology. No anomaly was observed on the face. Neurological and systemic examinations were normal, no arteriovenous malformation was detected. There was no other disease in the patient's history or family history. On hand X-ray, soft tissue enlargement of the left hand index finger and thumb phalanges and medial folding on the left hand index finger phalanges were observed (Figure 2). The patient was referred to Plastic and Reconstructive Surgery outpatient clinic for surgical treatment. Surgical treatment was recommended by Plastic and Reconstructive Surgery, but treatment could not be performed because the patient's family did not accept it.

Macrodactyly is a rare anomaly in which the fingers grow disproportionately and develop during birth or in the first years of life. In affected fingers, there is diffuse growth in soft tissue and bone. Most cases occur sporadically.



Figure 1. On physical examination, prominence of the left hand thumb and swelling and enlargement of the index finger and medial folding of the left hand index finger were observed

The anomaly is usually unilateral and if the thumb is affected, the probability of affecting more than one finger increases. The affected finger hardens during growth and enlarged fingers often obstruct movement (4).

Patients are generally evaluated for surgical indication for functional and aesthetic reasons. The management of macrodactyly is a challenging issue due to its progressive and widespread nature. Surgical options are limiting the ongoing growth, reducing the finger size or correcting the deviation and amputation (4). Macrodactyly may be alone or seen as a component of syndromes such as

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Proteus syndrome, Maffucci syndrome, Bannayan-Riley-Ruvalcabe syndrome, Ollier's disease, Milroy's disease, Klippel-Trenaunay-Weber syndrome. Therefore, patients with macrodactyly should be screened for accompanying syndromes (1,4). The patient did not have any pathological findings as a result of detailed eye examination and neurological examination and cranial magnetic resonance imaging. No hyperpigmented lesions, hemangioma or other vascular malformations, lipomatous lesions and hypertrophy of the lower extremities were detected in our patient.



Figure 2. On hand X-ray, soft tissue enlargement of the left hand index finger and thumb phalanges and medial folding on the left hand index finger phalanges were observed

Proteus syndrome is a syndrome that can show excessive growth in skeletal system, solid organs, skin and subcutaneous tissue. Especially, hypertrophy that can be seen in a single extremity or localized area, underlying hemangiomatous lesions, lipomatous lesions and hyperpigmented areas are important clinical features (2). Bever et al. and Guelmi et al. reported that isolated macrodactyly may be an extremely localized form of Proteus syndrome (5,6). Rubinstein-Taybi syndrome is a genetic disorder characterized by facial disorders, short stature, and moderate to severe intellectual disability, large hand and toes. In the first years of life, respiratory difficulties, nutritional problems, low weight gain, recurrent infections, and severe constipation may occur (7). Klippel-Trenaunay syndrome is a disease of unknown etiology

characterized by vascular malformations, varicose veins and asymmetric limb hypertrophy triad (8). Maffucci syndrome is a very rare congenital disease characterized by diffuse enchondroma and hemangiomas (3).

Ollier's disease is a non-hereditary mesodermal dysplasia characterized by numerous enchondromas with asymmetric involvement in the metaphyses of long bones. It is characterized by numerous enchondroma or displaced cartilaginous masses in the metaphysis and diaphysis of long bones in the hand bones (9). Milroy's disease is characterized by a congenital developmental anomaly of the lymphatic vessels resulting in edema starting from the dorsum of the foot and spreading to the lower extremities (10). No additional pathological findings were found in the patient who was hospitalized by Orthopedics and Traumatology, Neurology and Pediatrics. Dermatological examination did not show depigmentation. There was no characteristic facial appearance that could indicate mental disorder, height or weight abnormality or a syndrome. The patient was evaluated as isolated macrodactyly.

Macrodactyly is a very rare anomaly. We present this rare case with clinical image and plain radiography.

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