Fibrodysplasia ossificans progressiva (FOP) is an extremely rare disorder that is characterized by congenital skeletal malformations and by progressive heterotopic ossification of tendons, ligaments, fasciae, striated muscle and occasionally skin. Although the pattern of inheritance is autosomal dominant, most reported cases are the result of spontaneous mutations. There is no sexual, racial or ethnic predilection. Its incidence is reported to be approximately 1 in 2 million people. Heterotopic ossification usually begins in the cervical, occipital and upper paravertebral muscles and then involves muscles around all the major joints, producing progressive immobilization. At present, there is no effective prevention or treatment. Bone scintigraphy is effective for an early detection of heterotopic ossification and for monitoring the progression of the disease. We present a case of FOP who is an 28 years old female, with an interesting bone scan appearances.

**Key Words:** Fibrodysplasia ossificans progressiva, bone scintigraphy, Tc-99m MDP

**Abstract**

Fibrodysplasia ossificans progressiva (FOP) is an extremely rare disorder that is characterized by congenital skeletal malformations and by progressive heterotopic ossification of tendons, ligaments, fasciae, striated muscle and occasionally skin. Although the pattern of inheritance is autosomal dominant, most reported cases are the result of spontaneous mutations. There is no sexual, racial or ethnic predilection. Its incidence is reported to be approximately 1 in 2 million people. Heterotopic ossification usually begins in the cervical, occipital and upper paravertebral muscles and then involves muscles around all the major joints, producing progressive immobilization. At present, there is no effective prevention or treatment. Bone scintigraphy is effective for an early detection of heterotopic ossification and for monitoring the progression of the disease. We present a case of FOP who is an 28 years old female, with an interesting bone scan appearances.

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**Özet**


**Anahtar Kelimeler:** Fibrodisplazi ossifikans progressiva, kemik sintigrafisi, Tc-99m MDP

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present a case of FOP who is an 28 years old female, with an interesting bone scan appearances (Figure 1). The diagnosis of the patient is based on clinical and radiological findings and demonstration of skeletal malformations including bilateral malformed great toes and absent teeth. Briefly, there was cervical rigidity and a tendency to torticollis. There was contraction of the muscles of the pectoral girdle, especially in the right shoulder. Flexion, rotation and stretching of the nape were not possible, nor were movements of the trunk. Her brother and father had the same congenital deformity and probably, the disease was the autosomal dominant inheritance. Genetic linkage analysis have been planned.

REFERENCES