A Case of Brachymetatarsia From Medieval Sardinia (Italy)

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ABSTRACT

Archaeological excavations carried out in the Medieval village of Geridu (Sardinia) uncovered several burials dating to the late 13th or the first half of 14th century. Among these individuals, the skeleton of an adult female showing a bilateral abnormal shortness of the fourth metatarsal bone was identified. Bilaterality and absence of other skeletal anomalies allow to rule out an acquired aetiology of the disease and to support a diagnosis of congenital brachymetatarsia. Such a rare deformity has a clinical incidence of 0.02% to 0.05%, with strong predominance of the female gender. To our knowledge, no other cases of brachymetatarsia have been reported in paleopathology so far. Anat Rec, 00:000–000, 2014.

Key words: congenital diseases; shortened fourth metatarsal bone; Sardinia; Middle Ages

Modern medicine has classified a large number of congenital disorders involving defects in a developing fetuses and in infantile age; these disorders range from minor anomalies, generally asymptomatic, to the more severe abnormalities, some of which are incompatible with life. Although congenital defects have been reported in ancient human remains (Barnes, 1994; Ortner, 2003; Roberts and Manchester, 2007), it is still impossible to determine the range and incidence of congenital diseases in Antiquity.

Apart from attesting the presence of specific diseases in the past, these cases are of great topical interest. They allow to observe the evolution up to adult age of many defects most of which are corrected by surgery today; they also allow to infer the social behavior of past societies towards impaired or deformed individuals. All reports are important, considering the relatively small number of published archaeological cases of congenital diseases.

Here we report on a case of abnormal shortness of the fourth metatarsal bones, defined brachymetatarsia, from Medieval Sardinia.

Brachymetatarsia is a rare deformity, whose incidence in modern populations ranges from 0.02% (Urano et al., 1978) to 0.05% (Mah et al., 1983). The condition shows a

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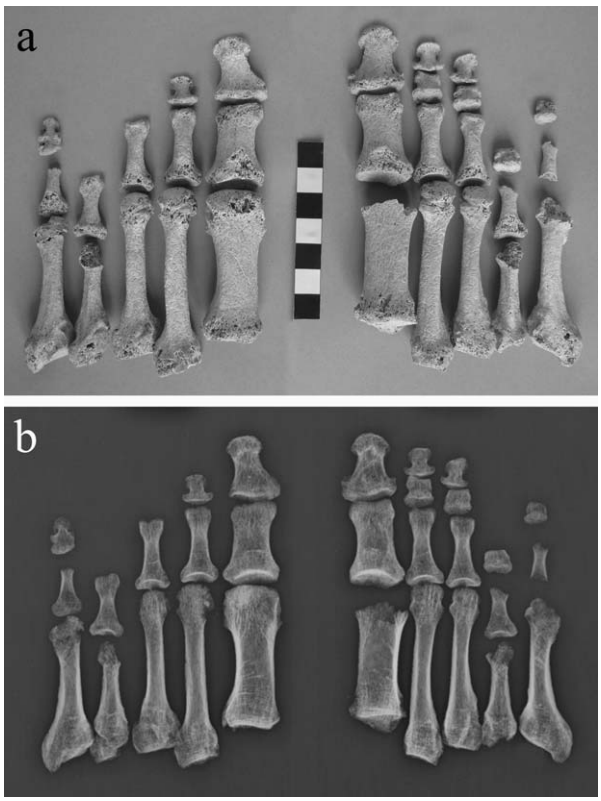

Fig. 1. Skeleton n° 2580 exhumed from the burial site of Geridu: the feet show abnormally shortened fourth metatarsal bones (**a**); AP projection of the feet (**b**).

strong female prevalence of 25:1 and the defect is bilateral in the majority of cases (72%) (Urano et al., 1978). The diagnosis of brachymetatarsia can be made when one metatarsal is 5 mm or more proximal to the parabolic arc of the metatarsal heads. Any metatarsal can be affected, but the fourth metatarsal results the most frequently involved (Schmizzi and Brage, 2004).

TABLE 1. Congenital syndromes and endocrinopathies that can cause brachymetatarsia (Schimizzi and Brage, 2004)

Congenital syndromes
Aarskog syndrome
Apert syndrome
Brachiodactyly type E
Carpenter's syndrome
De Lange syndrome
Down syndrome
Ectrodactyly
Grebe syndrome
Hypochondroplasia
Hajdu-Cheney syndrome
Hand-foot-genital syndrome
Jeune's thoracic dystrophy
Killian/Teschler-Nicola syndrome
Langer-Giedion syndrome
Leri-Weill dyschondrosteosis
Majewski type short rib–polydactyly
Mohr syndrome
Multiple synostosis syndrome
Multiple synostosis syndrome
Pfeiffer syndrome
Poland's anomaly
Rett syndrome
Robinow's syndrome
Rothmund-Thomson syndrome
Ruvalcaba's syndrome
Saethre-Chotzen syndrome
Taybi syndrome (Otopalatodigital syndrome)
Turner syndrome
Weill-Marchesani syndrome
Warfarin effects
Werner's syndrome X syndrome
18p syndrome
Endocrinopathies
Pseudohypoparathyroidism
Pseudopseudohypoparathyroidism
Diastrophic dysplasia
Multiple epiphyseal dysplasia
Myositis ossificans
Achondroplasia
Achrodysostosis
Acromesomelic dysplasia
Geleophysic dysplasia
Metaphyseal chondrodysplasia
Thanatophoric dysplasia

MATERIALS AND METHODS

The medieval village of Geridu, located in northern Sardinia, disappeared in the early decades of the 16th century and has been brought to light thanks to archaeological excavations carried out in recent decades. In particular, 25 single pit graves dating back to the late 13th or the first half of 14th century have been uncovered (Milanese, 2001; Milanese et al., 2004).

The skeleton of an adult individual (sample code number: 2580), showing abnormalities in the foot bones, was identified.

Sex determination was performed on the basis of the morphologic features of the skull (Ferembach et al., 1977–79; Buikstra and Ubelaker, 1994). Age at death was estimated on the basis of dental wear (Miles, 1963) and of sternal rib end modification (Loth and Iscan, 1989). Lesions indicative of pathologies were recorded in accordance with the methods and standards set out in the Global History of Health Project (Steckel et al., 2005).

Morphological macroscopic observation of the foot bones was followed by conventional X-rays. A FCR Velocity by Fujy direct DR equipment, with following parameters-10–12 mAs with 54–60 keV, DFF 110- cm, was used.

RESULTS

The skeletal remains belong to a female aged 35–45 years old.

The neurocranium is fairly well preserved, whereas the splanchnocranium is partially fragmented. The postcranial skeleton is represented by fragments of clavicles and *scapulae*, the left hand, the spine and the lower limbs; the remaining bones are missing. Level with both feet, some middle and distal phalanges are missing.

The paleopathological study revealed a Schmorl's node on a thoracic vertebra. Osteoarthritis of Grade 1 affected the thoracic vertebrae, and a Grade 1 degenerative joint disease (slight marginal lipping and slight degenerative/

productive changes present) was observed on the acromion-clavicular joint, distal femurs, articular surface for the talus of the calcanei and throclea of the talus bones. Diffuse periostitis of Grade 2 affected both tibiae.

The foot bones of the Geridu woman showed bilateral abnormal shortness of the fourth metatarsal bone; these bones appear about 2 cm shorter than the rule (Fig. 1a). Morphological macroscopic observation was followed by conventional X-rays (Fig. 1b). Fusion of the middle and distal phalanges of the left fifth toe is present. No evidence of other malformation was observed in the cranial and post-cranial skeleton.

DISCUSSION AND CONCLUSIONS

Brachymetatarsia is caused by premature arrest of the growth of the epiphysis and becomes apparent by approximately 10 years of age (Kim et al., 2003). There are three types of brachymetatarsia: idiopathic congenital, associated congenital and acquired (Munuera Martinez et al., 2004).

Several syndromes and disorders are at the origin of associated congenital brachymetatarsia (Table 1); acquired etiology refers to traumas, sickle cell crisis, juvenile rheumatoid arthritis, infection, tumor, and poliomyelitis (Schimizzi and Brage, 2004).

Patients with brachymetatarsia can suffer from metatarsalgia, calluses, shoe wear irritation, and soft tissue contractures. In addition, aesthetic problems accompany this condition. Nowadays, the defect is surgically corrected, by distraction osteogenesis using external fixation or with one-stage lengthening using bone graft (Giannini et al., 2010).

Since the defect observed in the Geridu woman is bilateral and is not apparently accompanied by further alteration of the remaining skeletal remains, this allows to rule out an acquired aetiology of the disease (i.e., traumatic) and to support a diagnosis of congenital brachymetatarsia. However, it is impossible to ascertain whether this is an isolated manifestation or whether it is part of a more complex syndrome, as the soft tissues have decomposed and several bones are missing. To our knowledge, no other cases of brachymetatarsia have been reported in ancient skeletal remains so far.

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