Musculoskeletal radiology

Osteoporosis – Pseudoglioma Syndrome: report of three brothers

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Abstract. Three sibs with osteoporosis-pseudoglioma syndrome (OPS) are described. Two older boys were diagnosed as osteogenesis imperfecta. In the younger boy OPS was recognised at first consultation. This syndrome radiologically simulates osteogenesis imperfecta. These two conditions can be easily differentiated if associated clinical and ophthalmological features are taken into consideration.

Key words: Osteoporosis – Pseudoglioma – Osteogenesis imperfecta

Introduction

Osteoporosis-pseudoglioma syndrome (OPS) is a rare but well-defined condition characterised by premature generalised osteoporosis and complex eye abnormalities. Approximately 40 cases had been reported up to 1993 [1–3, 5, 6, 9, 12–17]. In one Finnish family with two affected sibs several members of previous generations became blind and/or showed signs of bony involvement [5]. The purpose of this paper is to report three sibs with OPS in a Czech family (Table 1). This is the first report of OPS from the Czech Republic.

Table 1. Pedigree of the family with Osteoporosis-pseudoglioma syndrome. III 3 blindness, oligophrenia (not investigated); III 1 blindness, oligophrenia, multiple fractures; III 2 blindness, oligophrenia; III 3 healthy; III 4 proband: blindness, oligophrenia, congenital heart disease

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Case report

Case 1

The proband (TG) was born at 37 weeks of gestation weighing 2500 g and measuring 46 cm in length. He was the fifth child of a woman with sarcoidosis and her healthy husband. The mother's brother was mentally retarded and blind. He was not investigated. Two older brothers of the propositus were affected in a similar way (Cases 2 and 3; Table 1).

Upon examination at the age of 2 years his weight was 4520 g, height 65 cm and head-circumference 39.5 cm. He was microcephalic, hypotonic and showed severe psychomotor retardation. He had a ventricular septal defect and truncus arteriosus communis. An ophthalmological examination revealed micro-ophthalmia, microcornea, bilateral pseudoglioma, posterior-chamber synechiae and corneal dystrophy on the left.

Serum osteocalcin was low (11.8 μg/ml; normal values 25–30 μg/ml). Serum levels of parathormone, calcium, phosphorus and alkaline phosphatase were normal. A skeletal survey disclosed osteoporosis, but no de-
Fig. 1a–c. Case 1. Osteoporosis-pseudoglioma syndrome (OPS) a, b 6 days old. a Thin shafts. Slightly widened metaphyses. b Thin ribs. Bell-shaped thorax. c A 2-year-old. Short metacarpals. Bone age corresponds to the chronological age of 12 months.

formities or fractures (Fig. 1a–c). The OPS was diagnosed. He died at the age of 4 years following a respiratory tract infection. He had never been able to sit or stand.

Case 2

The oldest brother of TG was born after a normal full-term pregnancy and delivery. His birth weight was 3600 g and length 49 cm. An ophthalmological examination at the age of 7 months found that he was blind. He started to walk at the age of 15 months. Several fractures of his osteoporotic bones occurred during his second year of life with subsequent progressive deformities of the lower limbs. The diagnosis of osteogenesis imperfecta was made. He stopped walking at the age of 3 years and was bedridden until his death from a respiratory tract infection at the age of 14 years. He was mentally retarded less than his older brother, but he also could neither read nor write. Radiographic diagnosis of rickets at the age of 2 years was excluded as the serum calcium, phosphorus and alkaline phosphatase levels were normal. He suffered pathological fractures in his early school years and gradually developed progressive leg deformities. He became bedridden at the age of 10 years (Fig. 2a–g). Osteogenesis imperfecta was diagnosed, although Norrie-Wardburg syndrome was also suggested as a possible diagnosis. He died at the age of 20 years from a respiratory tract infection (Fig. 2H).

Discussion

The clinical signs and symptoms of OPS are variable. Fractures with bony deformities and progressive loss of vision are the major diagnostic criteria. Osteoporosis is a constant radiographic finding, whereas the number of fractures and deformities are variable. The lower extremities and the spine are most often affected. Wormian bones are often present, and it is not surprising that the radiographic diagnosis of osteogenesis imperfecta is often made, especially if the clinical findings

Case 3

The second brother of TG was born after a normal full-term pregnancy and delivery. His birth weight was 3800 g and length 51 cm. Blindness was recognised in his first year of life, but until his death he retained the perception of light. His motor development was minimally delayed, with sitting at 7 months, standing at 14 months and walking at 15 months. He was mentally retarded less than his older brother, but he also could neither read nor write. Radiographic diagnosis of rickets at the age of 2 years was excluded as the serum calcium, phosphorus and alkaline phosphatase levels were normal. He suffered pathological fractures in his early school years and gradually developed progressive leg deformities. He became bedridden at the age of 10 years (Fig. 2a–g). Osteogenesis imperfecta was diagnosed, although Norrie-Wardburg syndrome was also suggested as a possible diagnosis. He died at the age of 20 years from a respiratory tract infection (Fig. 2H).

Fig. 2a–h. Case 3. OPS a–h A 13-year-old. a, b Thin long bones. c Small cranium in relation to the face. d Thin ribs. Bilateral inflammatory changes. e Extremely thin, deformed tibia and fibula. f Narrowing of the joint spaces. g Irregular, mild platyspondyilia of the thoracic spine. h A 20-year-old. Microcranium. Severe deformities of the chest, spine and lower extremities.