Case Report

A case report of late diagnosed osteogenesis imperfecta

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Abstract

Osteogenesis imperfecta (OI) is a rare and inherited disorder that causes a generalized decrease in bone mass and makes the bone brittle. The disorder is frequently associated with blue sclera, dental abnormalities, progressive hearing loss, and a positive family history. The most severe forms of OI may cause in-utero death, still-birth or death shortly after birth. The course of mild and moderate forms is more variable. Some patients may appear normal at birth and worsen progressively. Some may have multiple fractures in infancy and childhood; improve after puberty, and have fractures more frequently later in life. Genetic and prenatal counseling, screening, and close follow-up of the family members are essential issues for primary care physician. A 38-year old woman admitted to our clinic for low back pain. Her examination revealed blue sclera, temporamandibular joint subluxation and scoliosis. Her dentition was all prosthesis. In the family history, her three of five children, her sister, brother, mother and grandmother had blue sclera and her children had spontaneous fracture history and also her brother had hip prosthesis surgery. She was diagnosed OI. Her management was planned and performed in multidisciplinary approach to treat the existing symptoms and signs, to prevent complications and to improve the quality of life. Three of her children were consulted to pediatrician and diagnosed OI. This case report emphasizes the importance of early diagnosis of hereditary diseases and role of a regular family physician providing continual care in early diagnosis and management of such inherited disorders.

Key Words: Osteogenesis imperfecta, adult, primary care

Sezer T, Ozcan S, Saatci E. A case report of late diagnosed osteogenesis imperfecta. TJFMPC, 2008;2(4):56-59.

Introduction

Osteogenesis imperfecta is an inherited disorder that causes a generalized decrease in bone mass (osteopenia) and makes the bone brittle. The disorder is frequently associated with blue sclera, dental abnormalities (dentinogenesis imperfecta), progressive hearing loss, and a positive family history.[1] The other clinical features are growth deficiency, skin thinness, joint laxity and hipermobility.[2-4] Molecular studies have demonstrated that most cases result from mutations affecting the two genes (COL1A1 and COL1A2) responsible for the formation of type 1 collagen.[5,6]

The most common classification for osteogenesis imperfecta was developed by Sillence[7]. Type I, the mildest form, is inherited as an autosomal dominant trait. Most patients have distinctly blue sclera. Type I is subdivided into types IA and IB depending on whether or not dentinogenesis imperfecta is present. Type II is lethal in utero or shortly after birth. Type III and IV osteogenesis imperfecta are intermediate in severity between types I and II. They differ from type I because of lesser severity and because the sclera are only slightly bluish in infancy and white in adulthood. Type III differs from type IV in that it tends to become more severe with age. It may be sufficient to classify patients simply as mild (type I), lethal (type II) and moderately severe (type III). Type I has a frequency of about 1 in 30.0000. The incidence of Type II osteogenesis imperfecta at birth is 1 in 60.000, but the incidence of the three severe forms recognizable at birth (type II, III and IV) may be as high as 1 in 20.000.[1] Genetic and prenatal counseling, screening, and close follow-up of the family members are essential issues for primary care physician.[8]

We present a late diagnosed osteogenesis imperfecta case to emphasize the role of family physician and the importance of continuity of care in early diagnosis and management of such inherited disorders. The assurance of anonymity was provided and written consent was obtained from the patient.

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Submitted date: .19.09.2008
Accepted date: 02.11.2008

CASE DESCRIPTION

A 38-year-old woman admitted to our outpatient clinic for low back pain. There was no history of trauma. In her medical history, she had several visits to different specialists (such as orthopedist, dentist, neurologist and gynecologist) for low back pain, dental problems, headache and abdominal pain. She had bilateral progressive hearing loss since her childhood. In physical examination; her height was 1.45 m, body weight: 51 kg, body mass index (BMI): 24.2, blood pressure: 140/70 mmHg. She had blue sclera and her dentition was all prosthesis. Musculoskeletal examination demonstrated scoliosis and temporamandibular joint subluxation.

Her family history revealed migration from East of Turkey. The socioeconomic status was low and they had language problems. Her three of five children, her sister, brother, mother and grandmother had blue sclera. Her son and daughter had history of spontaneous femur fracture; her brother had hip prosthesis surgery (Figure 1).

Complete blood cell count and blood biochemistry tests were normal. Radiographic evaluation showed flattening of the normal curve of lumbar vertebra and scoliosis with right-sided curve, narrowing in L2-3 on the right-side and in L5-S1 on the left-side, sclerosis and degenerative osteophytes. Height of vertebra corposis was normal (Figure 2).

Our diagnosis in this patient was osteogenesis imperfecta type I because of decreased mineral density, slightly bluish sclera, prosthesis of all teeth (probably due to dentinogenesis imperfecta), extra-skeletal abnormalities (scoliosis and temporamandibular joint subluxation) and positive family history of the blue sclera and fractures. She was consulted to orthopedist, physical therapy and rehabilitation specialist and ear, nose and throat specialist. Her dual-energy x-ray absorptiometry (DEXA) revealed osteoporosis (T score L3: -3.1, Z score: -2.9). Physical therapy and medical therapy for osteoporosis was planned. Audiogram was planned and bilateral sensorineural mixed type (right ear 61dB, left ear 66 dB) hearing loss was found. Explorative operation for the left ear and hearing aid were planned. The patient and her husband were informed about the disease and were given genetic counseling. Their children were screened. Three of them were affected. They were consulted to the pediatrician. Our diagnosis was confirmed and alendronat 10 mg/day calcium carbonate 2500 mg/day, vitamin D3 880 UI/day was prescribed for osteoporosis.

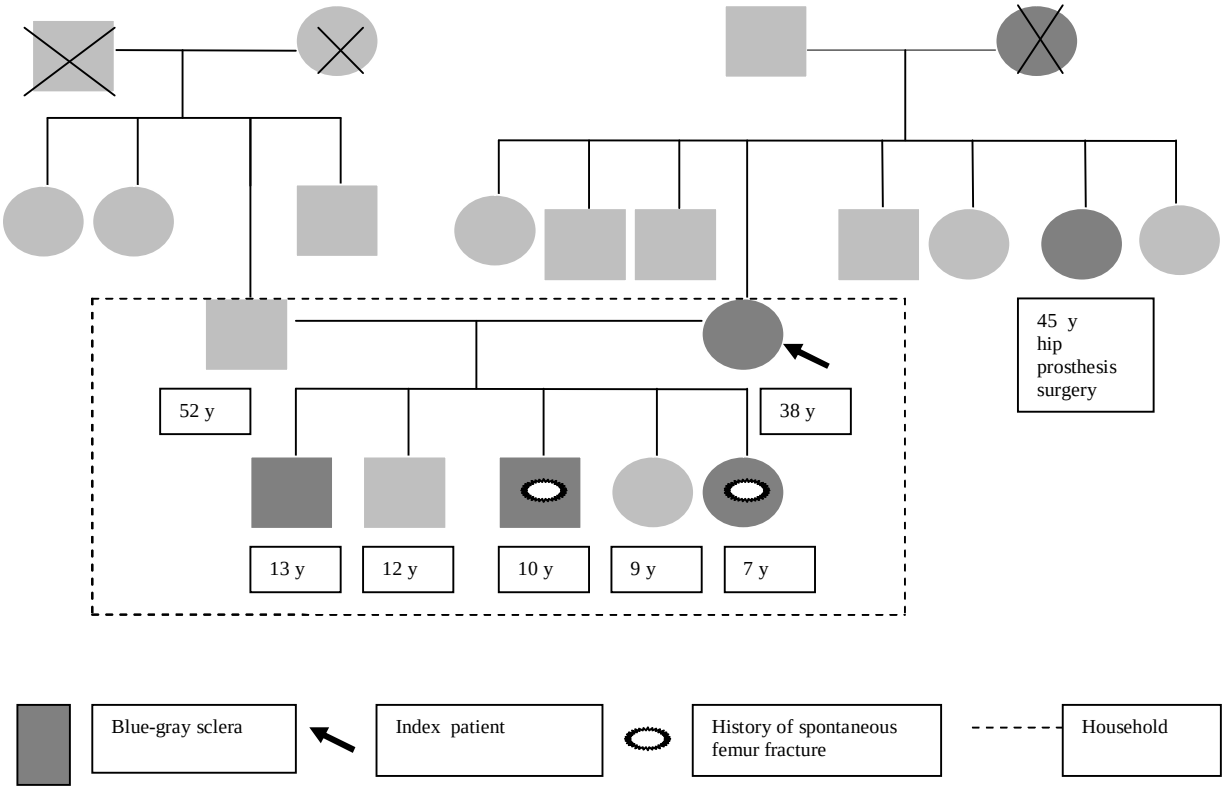
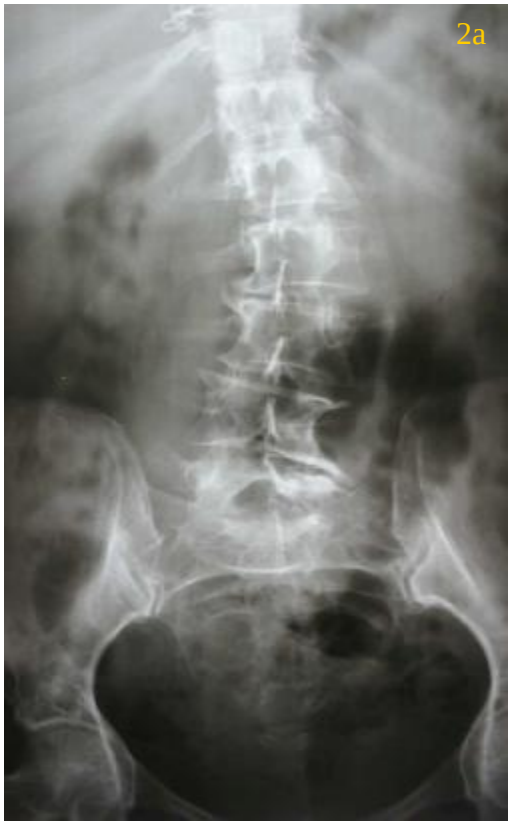


Figure 1-Family pedigree



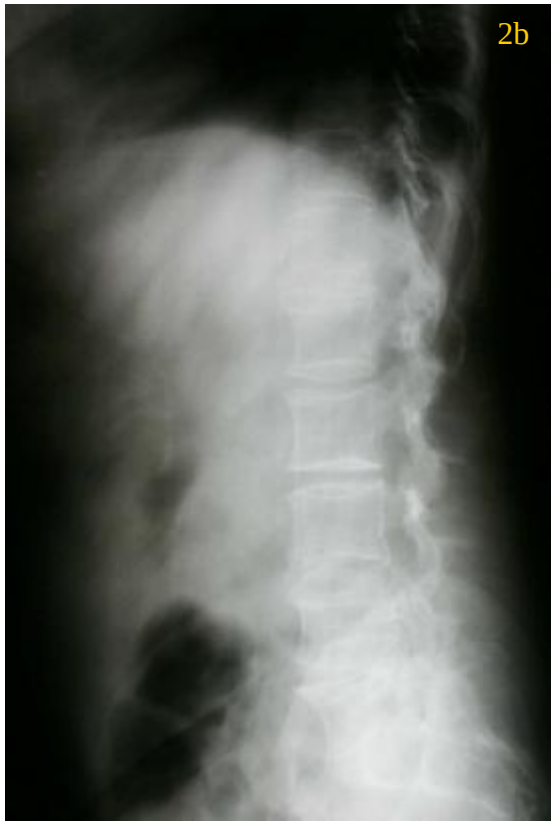


Figure 2a and 2b- Anteroposterior and lateral lumbosacral X-rays showing flattening of the normal curve of lumbar vertebra and scoliosis with right-sided curve, narrowing in L2-3 on the right-side and in L5-S1 on the left-side, sclerosis and degenerative osteophytes.

Discussion

The most severe forms of osteogenesis imperfecta may cause in-utero death, still-birth or death shortly after birth. The course of mild and moderate forms is more variable. Some patients may appear normal at birth and worsen progressively. Some may have multiple fractures in infancy and childhood; improve after puberty, and have fractures more frequently later in life. Women are particularly prone to fracture during pregnancy and after menopause. A few women from families with mild variants of osteogenesis imperfecta do not develop fractures until after menopause and their disease may be difficult to distinguish from postmenopausal osteoporosis.[1]

The diagnosis of osteogenesis imperfecta is usually made on the basis of clinical criteria. The presence of fractures together with blue sclera, dentinogenesis imperfecta or positive family history are usually sufficient to make the diagnosis, but can be difficult in the absence of affected family members and when bone fragility is not associated with obvious extra-skeletal abnormalities.[1]

Blue sclera may be the first sign suggestive of a diagnosis of osteogenesis imperfecta. It can be normal, slightly bluish or bright blue. The teeth may be normal, moderately discolored or grossly abnormal. The enamel generally appears normal, but the teeth may have characteristic amber, yellowish brown or translucent bluish-gray color due to improper deposition or deficiency of dentin. Hearing loss usually begins during the second decade of life and occurs in more than 50% of subjects over age 30. The loss can be conductive, sensorineural or mixed and may vary in severity.

In all forms of osteogenesis imperfecta, bone mineral density is decreased.[1] Scoliosis is common. The combination of chest deformities and scoliosis is responsible for a large number of deaths due to the respiratory disorders.[10,11] Ligaments have high type I collagen content so that ligamentous laxity is one of the features.[12,13] Hypermobility as a cardinal feature of osteogenesis imperfecta may present as

arthralgias and dislocations. Lax joints are likely to be less stable, to subluxate or dislocate. Soft tissues are less resilient so that ligament and muscle tears and tendon osseous attachment lesions may occur with increased frequency.[13]

Patients with mild disorder may need little treatment when fractures decrease after puberty, but women require special attention during pregnancy and after menopause when fractures increase again. More severely affected children require comprehensive program of physical therapy, surgical management of fractures and skeletal deformities and vocational education.[1]

Early diagnosis of this disease will be an initiation of therapy as well as patient education regarding management of modifiable risk factors linked with osteoporosis. [8] Family screening should also be offered to families and, genetic counseling and emotional support are important for patients and parents. Prenatal ultrasonography will detect severely affected fetuses at about 16 weeks of pregnancy. [1]

The management of patients with osteogenesis imperfecta requires the involvement of a multidisciplinary team and a family physician is the essential professional for coordination in these patients. In Turkey, a patient can admit to primary, secondary or tertiary care without consulting any gate-keeper. Our patient had admitted to several specialists. It is unknown whether a diagnosis of OI was thought by the specialists as neither she nor her non-existing medical records told so. Another probability is that the patient did not understand or recall the given medical information due to her low educational status, language problem and hearing loss. This missed diagnosis emphasizes the role of a personal family physician providing continual care in early diagnosis and management of such inherited disorders.

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