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Osteogenesis imperfecta associated with recurrent depressive episodes and postpartum psychosis in a 27-year-old women

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Summary

A 27-year-old woman was admitted to a psychiatric ward due to acute postpartum psychosis. The patient suffered from both recurrent depressive episodes with histrionic traits for years and osteogenesis imperfecta type I according to Sillence. As the patient and her monozygotic twin, also afflicted with comparable psychiatric symptoms, were the first combined psycho-osteological disorder of monozygotic twins. The patient recovered within a few weeks following antipsychotic and antidepressant treatment.

Osteogenesis imperfect / recurrent depression / postpartum psychosis / osteology / chromosome 17 / gene

INTRODUCTION

Osteogenesis imperfecta (OI, brittle bone disease) is an autosomal dominant bone disease, but recessive entities and spontaneous forms occur. The prevalence is approximately 3-7 per 100000 [1, 2]. The pathobiochemistry is characterized by substitution of glycine by larger amino acids leading to an impaired triple-helix structure of collagen fibrils composed of 3 tropocollagen chains each, type I collagen being the most important protein of the bone matrix [3]. This deficiency leads to an improper integration of hydroxyapatite, which may be responsible for the semiology of the connective tissue disease [4].

Nowadays at least eight types of Osteogenesis imperfecta have been identified and it is not the purpose of this case report to go into pathohistological details [1]. Particularly lack or deficient type I collagen increases the risk of fractures and deformations bones, joints and spine. Other characteristical symptoms are blue sclerae, loss of hearing, slight protrusion of eyes, short stature, valvular heart disease, hernias or impaired vision [5, 6]. However, the severity of the disorder may vary extremely, type I is rather mild whereas type II can even cause intrauterine death. The most important genetic cause of OI is a mutation on chromosome 17 (Col 1A gene), which codes for the 2 pro-alpha1-chains composing the triple-helix together with a 1 pro-alpha2-chain (Col 2A gene) coded on chromosome 2 [7]. Particularly, in Osteogenessis type I the Col1A is involved and the autosomal dominant disease shows de novo mutations in approximately 60% [8]. The long arm of chromosome 17 is also coding for the serotonin transporter Gene (positions 17q11.1–q12 versus 21.3 and 22.1 Col gene), which is discussed as a genetic base of depression [9].

Anecdotal possible associations between osteogenesis imperfecta or bone deformations and psychiatric disorders have been reported very rarely since 1950 [10, 11, 12]. Chodirker and Var-

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samis [13] published two cases of schizophrenia in one family associated with osteogenesis imperfecta. Heide [14] lineated a complex syndrome consisting of osteogenesis imperfecta, mental retardation, bone deformations, hyperextensible joints and blindness in 3 siblings. The aim of this case report is not to consider sequelae of treatment with antidepressant or antipsychotics on bone mass [15], neither adjustment disorders due to lack of coping skills. On the contrary, the case report should emphasize the possible relation between congenital osteogenesis imperfecta and the potential associated disposition for affective and psychotic disorders.

CASE REPORT

A 27-year-old woman was admitted to the department of psychiatry due to acute psychosis by an emergency physician. She had developed severe – unrealistic – bizarre ideas of conspiracy involving mainly her mother-in-law and uttered diffuse threats towards her family. Intermittently, she heard voices and was convinced to be able to predict actions of her relatives.

She had delivered a boy two weeks before and had transiently been treated in an external psychiatric department for postpartum psychosis with antipsychotics and then parented at home, her child being placed in a foster family. According to their information, pregnancy was overall good. The delivery was by caesarean section and about 12 weeks before the actual date of birth. The postpartum wound healing process was uncomplicated. The lactation was stopped with a dopamine agonist bromocriptine.

Simultaneously, her husband had been admitted to a university hospital because of an exacerbated schizophrenia, which also could have triggered the depressive component of the disease due to marked emotional stress. The patient lived until the birth of the child and the exacerbation of the schizophrenic illness of her husband own household of around 60 km from the city. The contact with the parents was rather of reduced nature. Regular contact was with the sister and their companion. The patient was professionally trained and worked as an office clerk. Beforehand, the patient had been treated in our department several times due to recurrent depressive episodes (ICD 10: F33.1), temporarily combined with histrionic traits. The medical history also confirmed a congenital osteogenesis imperfecta (ICD 10: Q78.0) Type I according to Sillence [Col1A1 gene] and hyperlipoproteinemia (ICD 10: E78.2) [16]. Deformations of the spine and pelvis are shown in Fig. 1. After she had recovered, she gave informed consent to publish her case.

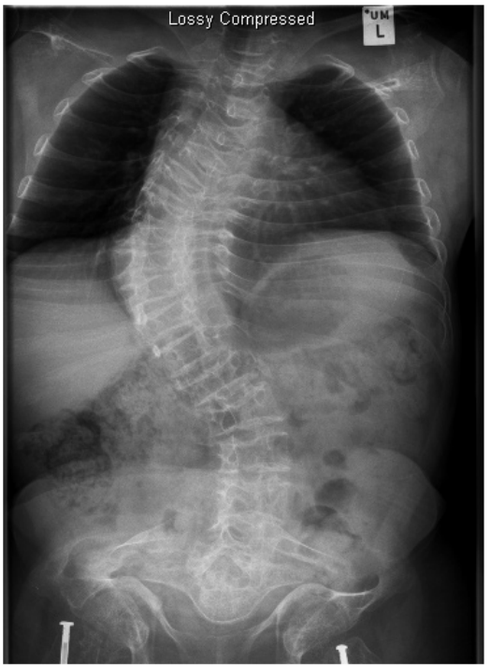

Figure 1. X-ray of the spine showing the marked pelvic and scoliotic deformation

CLINICAL EXAMINATION ON ADMISSION

Somatic State

The patient presented was in good nutritional condition but in slightly reduced general state of health (Weight: 39.8 kg / 87.744 pounds or lbs; Height: 110 cm / 43.307 inch). She was vertically challenged with obvious skeletal deformations but painless mobility. Auscultation, percussion and palpation of lung, heart and abdomen were normal. No skin or mucosal abnormality was detected. The cicatrice of the caesarian healed well. No neurological deficit could be ascertained.

Mental State

The patient was alert and conscious with normal orientation. Dress and appearance were ad-

equate, but with regard to communication she was cautious, ambivalent and suspicious. Her behavior was bizarre and peculiar with mannered facial expression and gesture. She spoke fluently. She appeared agitated with increased drive. No cognitive or mnestic deficit could be ascertained. Thoughts were erratic and accelerated with asyndesis, partially thought broadcasting and paranoid ideas (of persuit, threat and reference). Acoustic hallucinations (voices) were reported without any hint for impaired self-awareness. Mood was elevated but instable with reduced resonance. She was not anxious, nor did she complain of obsessions or hypochondria. She expressed no insight but was willed to be treated on the wards. Sleep quality was impaired with reduced period of sleep. She had no pain or additional autonomous symptoms. The ability to critically judge was reduced.

Family history

The (monozygotic) twin sister of the patient also suffered from osteogenesis imperfecta and had been treated in psychiatric hospitals by reason of acute transient psychotic disorders which were complicated by a possible histrionic personality disorder. Depressive episodes with suicidal thoughts and anxiety had been documented during adolescence. The 70-year-old mother suffered from poliomyelitis but has had no psychiatric disorder so far.

Supplementary tests and investigations

Blood pressure and ECG were normal on admission (BP: 120/75 mmHg, HR 72 bpm, intermediate cardiac vector without conductions abnormalities). Clinical laboratory work up showed normal values (SI units) apart from LDL (4.07) and hemoglobin (7.0)

Due to the recent delivery gynecological consultant assessed the status with pathological findings. The EEG revealed an alpha-rhythm with slow activity without focus or epileptic patterns.

TREATMENT AND COURSE

She was treated with risperidone (1mg bid), mirtazapine (15mg hs) and lamotrigine was started. Ablactation was completed with carbergoline. The patient was enclosed in a multiprofessional and multidimensional therapeutic setting. She wished guardianship and a power of attorney was applied for, also with regard to her complex psychosocial relations. She recovered slowly within three weeks and could be discharged from the ward following another three weeks of stabilization including increasing exposure to daily routine. She could be entrusted in her parents care in good health, sufficient activities of daily living and free of psychotic symptoms. Unfortunately, she relapsed twice during the following four months and was re-admitted to the intensive care unit of the psychiatric department. Risperidone was replaced by quetiapine and lamotrigine was initiated as a maintenance drug with regard to future episodes. She responded within a few weeks and the medication is well tolerated. We are now looking for a suitable long-term accommodation for the patient. She was readmitted 3 months later due to suicide attempt (intoxication with different drugs) due a psychosocial conflict constellation. After intensive care monitoring drug doses were adjusted and she recovered with 2 to 3 weeks. A residential home was organized in order to stabilize the social setting.

DISCUSSION

We presented the first case of monozygotic twins who suffered from osteogenesis imperfecta, type I according to Sillence, and concomitant recurrent depression with psychotic episodes and histrionic traits. No bone disease or psychiatric disorder was found in the family that could be ascertained. As the responsible Col1A1 gene is located on the long (q) arm of chromosome 17 between positions 21.3 up to 22.1 [17]. Interestingly, the serotonin transporter gene between locations 17q11.1 and q12 [18], which is associated with depression, we speculate about a co-expression of the genes in our siblings.

The patients were treated according to current guidelines and recovered from acute depressive and psychotic symptoms within reasonable time. We are quite aware of the fact that psychiatric treatment is a risk factor for bone disease, even though this risk may be low. Regular consultan-

cy by and cooperation with an experienced osteologist seems to be absolutely mandatory.

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