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CASE REPORT

Rare Association of Schizophrenia in a Patient with Multiple Exostoses

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ABSTRACT

We report the case of a patient who suffered from schizophrenia with multiple exostoses (osteochondromatosis) and admitted in our psychiatric ward and argue about the possible role of exostosin (EXT) gene and its nearly chromosomal loci in further genetic studies of schizophrenia.

Only three cases have been reported till date which tried to associate coexistence of multiple exostoses and schizophrenia, thereby guiding further studies in genetic etiology of schizophrenia.

Keywords: Genetics, Multiple exostoses, Osteochondromatosis, Schizophrenia.

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CASE REPORT

A 22-year-old graduate unmarried female from urban setting was brought to the psychiatry outpatient department in January 2017 with 2 months duration of social withdrawal, persecutory delusions, third-person auditory hallucination of derogatory type, disorganized speech off and on, restricted mood and affect with lack of insight, and impaired sleep and personal hygiene. Onset was gradual with waxing and waning pattern for 2 years. She was previously prescribed antipsychotic medications and she responded to the treatment but discontinued after 8 months by herself. However, she never attained her premorbid level of functioning. There was no positive family history of psychosis. She was diagnosed having schizophrenia of paranoid type (F20.0 of the International Classification of Diseases, Tenth Edition, ICD-10).

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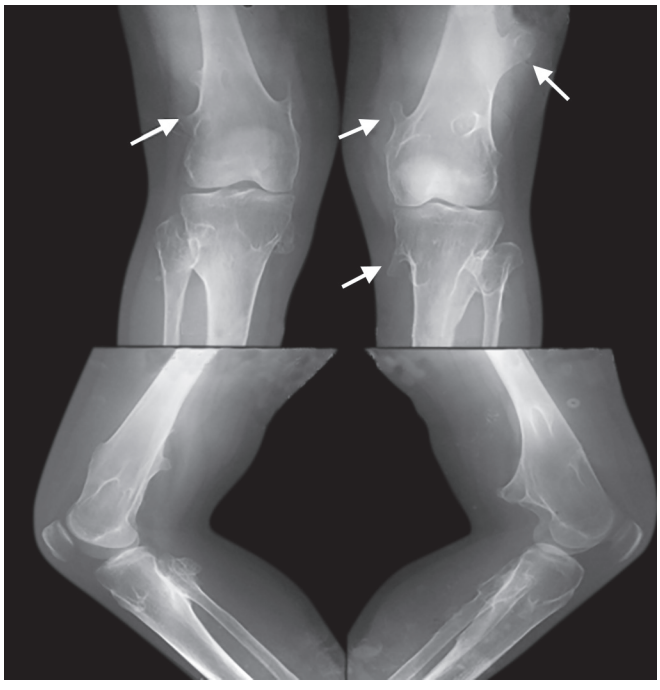

Fig. 1: X-ray of the patient showing multiple exostoses in both knee joints

General blood chemistry was within normal limits except low vitamin D_3 (17.9 ng/mL). Noncontrast computed tomography brain and electroencephalogram were also found to be normal. General and systemic examination did not reveal any significant finding. She was managed with Inj. Haloperidol and then later discharged with Tab. Olanzapine 10 mg/day.

She was also diagnosed having multiple exostoses in March 2016 for which she was operated on her right knee joint 7 months previously. However, there was no family history for the same. X-ray showed multiple exostoses over bilateral knee joints which was also confirmed by postoperative biopsy (Fig. 1).

DISCUSSION

Osteochondromatosis or multiple exostoses are benign cartilage-capped bone tumors that grow outward from metaphyses of long bones. Median age of onset is 3 years, nearly all affected are diagnosed by 10 to 12 years with autosomal dominant inheritance. Gene mutations responsible for it are EXT1 (8q24.11), EXT2 (11p11.2), and EXT3 (Chr 9). Penetrance is reported to be 96% in females

and 100% in males. However, nearly 20% cases have no positive family history.[1]

Age of onset (around 20 years) and psychiatric manifestations like paranoid delusions, incongruent and restricted mood and affect, lack of insight, disorganized behavior of our patient were comparable with those of Aizenberg et al[2] and Bernal[3]. Good response to antipsychotic drugs in controlling psychotic features was also comparable to that of Aizenberg et al[2] case reports, while Bernal[3] reported poor response to neuroleptics in his case. In all the three previously reported cases, surgical excision of exostoses was done, as similar to our case. Our case also does not have any positive family history of multiple exostoses.

Exostoses are rarely present at birth, but gradually arise and increase in size with age, with a wide spectrum of clinical presentation from only radiologically distinguishable signs to different skeletal deformities that remain minor physical anomalies of schizophrenics.[4]

Genetic transmission, correlation of phenotype and genotype, and variable spectrum of clinical manifestation are comparable in both the condition of schizophrenia and multiple exostoses. All the shared features and the cases reported earlier guide us to believe in the important role of EXT genes and nearly chromosomal loci in further studies in the genetics of schizophrenia.

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