Acute dystonia in a patient with 22q11.2 deletion syndrome

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Case Report

We describe the case of an 18-year old patient suffering from 22q11.2 deletion syndrome. Since adolescence, he manifested with behavioral disorders, aggression, verbal abuse and sleep disorders. Diagnosis of 22q11.2 was confirmed at the age of 3 with a special chromosomal detector (D22 s75, ONCOR). His performance at school was poor. After a heated argument with his parents, he presented intense anger, started breaking and throwing things and was verbally aggressive. His parents contacted the treating physician, who recommended treatment with 2 mg alogperidin. The patient received treatment dissolved in water. The treatment calmed him down, but a few hours later, he presented with cervical dystonia and emergence of torticollis and trunk dystonia with continuous twisting movements of the trunk and limbs, as well as accompanying dysarthria and pain. The patient visited the emergency outpatient clinic of the hospital, where he was given 1 amp Biperiden i.m. His acute dystonia was considered a side effect of alogperidin administration and he was subsequently subjected to full laboratory testing to investigate the risk factors associated with acute dystonia. The brain magnetic resonance imaging (MRI) revealed no abnormal findings and he was then subjected to a DATscan to investigate a possible dysfunction of the basal ganglia, which proved negative. After two weeks the patient was hospitalized in a psychiatric clinic and was administered quetiapine 200 mg, p.o. This particular treatment improved his clinical picture, without manifesting the usually induced side effects evoked by haloperidol.

Discussion

A literature search was conducted using the Medline computer database. It focused on all studies concerning 22q11.2 deletion syndrome and dystonia. The keywords were selected from titles, abstract and keywords and they were the following: Dystonia parkinsonism and di George syndrome. Butcher et al4 suggest that 22q11.2 deletions represent a novel genetic risk factor for early-onset of Parkinson disease (PD) with variable neuropathological presentation reminiscent of LRRK2-associated PD neuropathy. Shulman et al5 emphasize that the 22q11.2DS-associated microdeletion affects approximately 30 genes, including several intriguing candidates with potential links to PD pathophysiology. Among the genes in this region is COMT, encoding a key enzyme involved in dopamine catabolism and an established therapeutic target in PD. Zaleski et al6 suggest that the genetic of PD is complex. An increased risk to first degree relatives, even in apparently sporadic cases, suggests multifactorial causation of PD. Hereditary forms account for only about 1-3% of all PD cases and tend to have younger age at onset. Mutations in parkin, PINK1 and DJ1 are known to cause autosomal recessive early onset PD and mutations in alpha synuclein, ubiquitin C terminal hydrolase and LRRK2 can cause autosomal dominant PD with variable age of onset. Pinquier et al7 report a case of 22q11 deletion in a 17-year-old girl that was initially diagnosed as paranoid schizophrenia. In the case describe, transitory hypocalcemia induced dystonic symptoms that were believed to be cata
tonic symptoms or neuroleptic secondary effects. Verri et al8 describe a new male patient, 33-year-old, with 10p partial deletion syndrome and a severe autistic syndrome associated with mental retardation. The patient presented with dysmorphic features and at neurological evaluation, bilateral complete cataract, hypotonia, symptoms have been observed in 14% to 28% of children with di George syndrome and the receipt of antipsychotic treatment increase the risk of dystonic symptoms.9 Dystonia induced by alogperidin may arise from an imbalance between dopaminergic and mus-
Carinic receptors activity in nigrostriatum in the brain. The possible side effects of antipsychotic medications—either accelerating or revealing dystonic symptoms—merit further study.

Conclusions

Studies of di George syndrome add to the global understanding of the pathophysiology of the neurological symptoms of this common neurodegenerative disease.

References