

Psychotic Episodes in Adolescent with 22q11.2 microdeletion syndrome and underdiagnosed autism spectrum disorder

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Microdeletion 22q11.2 syndrome (22q11.2DS) is the most common microdeletion syndrome and encompasses the phenotype described as DiGeorge syndrome. It has a prevalence of 14.1:100,000 and the main clinical features includes thymic hypoplasia, hypocalcaemia, outflow tract defects of the heart, and dysmorphic facies. Eighty-four percent of the children with 22q11DS had at least one psychiatric disorder, including anxiety disorders, attention deficit [hyperactivity disorder](#) and about 16% met strict criteria for autism spectrum disorder (ASD).

We report a female patient with 22q11.2DS and underdiagnosed autism spectrum disorder evaluated in psychiatric consultation for the first time for inaugural psychotic symptomatology at 15 years old. She had learning difficulties with limitations in abstraction, planning, writing and attention deficit, in childhood. All these symptoms were interpreted as developmental delay and attention deficit hyperactivity disorder related to 22q11.2DS. At 15 years old, she began to present psychotic symptoms and was referred to a psychiatric emergency service, and she was hospitalized for 15 days, medicated with aripiprazole 5-10mg and clonazepam 2mg. When she went to the [psychiatric consultation](#), after this psychotic episode it was possible to understand that she always had social impairment, restricted interests and routines, good visual memory, and high sensitivity to noise, since childhood. After revision of all clinical data, we concluded that DSM 5 criteria for ASD were present. Diagnosis was confirmed with Autism Diagnostic Interview Revised (ADI-R).

Discussion:

This paper aims to alert to the possibility of confluence of 22q11.2DS with autism and psychotic symptoms and the possibility of [misdiagnosis](#). Sometimes autism spectrum disease symptoms are not diagnosed and are interpreted as cognitive delay. It is important to get a complete clinical report, and it is useful the parents 'contribute. We want to stress the importance of a multidisciplinary team, including the evaluation by a child psychiatrist, in developmental disorders.

Biography

Alda Mira Coelho -Child Psychiatrist / Child and Adolescent Psychiatry Service in Hospital S. João. Invited Assistant of Child and Adolescent Psychiatry -Faculty of Medicine, University of Porto /Master's degree in Developmental Psychology and Early Intervention, in Autistic children, in at the Faculty of Psychology and Educational Sciences and Submitted PhD thesis in the area of autism spectrum disorders: Prognostic Factors, in Faculty of Medicine, Univ. of Porto. She contributed to develop an integrated intervention project in the area of Autism, with specialized support units for ASD, in public schools, with an integrated psychoeducational approach, in addition to training actions and participation in scientific meetings. She developed a Specialized Consultation on Autism Spectrum Disorders (ASD), having carried out numerous training and support actions in this area, enhancing the importance of parents' in ASD.

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