Features of Autism in a Singaporean Child with Down Syndrome

Dear Editor,

We would like to present a case report on a 10-year-old Singaporean boy, John (name has been changed to preserve patient's confidentiality), diagnosed with Down syndrome (DS) (nondysjunction 47, XY+21) at birth and subsequently displaying autism-like behaviours. He was diagnosed with global development delay and attended special education programmes since 5 years of age. His parents and siblings displayed typical development. At 10 years 11 months, John underwent a comprehensive clinical assessment in view of concerns of autism-like behaviours. He was assessed on: (i) Autism Diagnostic Interview-Revised (ADI-R);[1] (ii) Autism Diagnostic Observation Schedule (ADOS);[2] (iii) Vineland Adaptive Behavior Scale (VABS);[3] and (iv) Aberrant Behavior Checklist (ABC).[4] In addition, results from blood tests revealed that he had normal thyroid function.

Case Report

Developmental History (0 to 4 Years)

John was born full term via normal vaginal delivery. He developed typically for a child with DS for the first 4 years. He used single words such as "Mummy" and "purple" at 2 years of age. He was socially responsive and had appropriate eye contact. He responded appropriately when asked "What's your name?" and "How old are you?" At 3 years of age, he was able to play functionally with a toy telephone. He engaged in reciprocal play by rolling toy cars to and fro with others. He participated in spontaneous pretend play by offering his mother a cup of drink and saying "for you". He spontaneously imitated actions, such as his parents snoring and penguins walking. He walked independently at 39 months.

John attended preschool at 4 years of age. He could follow simple instructions. For example, he was able to get his school bag from the locker, wipe the table, and put away his work. He needed individual supervision for table work such as pouring beans, cutting fruit, and shape puzzles. He used short phrases (e.g. "Mum scared" and "Stop it"). He was socially responsive to other children. However, some of his social overtures were inappropriate, such as hitting other children or pulling their hair.

Regression (4 to 7 Years)

John regressed at about 4 years 1 month. He lost communicative intent and the use of words that he had previously mastered. He showed no interest in toys and play. His eye contact became inconsistent and he was socially unresponsive. He rarely participated in group activities and had difficulty following instructions. He also started to display repetitive stereotyped behaviours such as rocking his body, flapping, and flicking his fingers. Several psychosocial stressors occurred around the time of John's regression, including the birth of his younger brother (at 3 years 10 months), his elder sister starting formal schooling (at 4 years 2 months), and the family moving house (at 4 years 9 months).

7 Years to 10 Years 11 Months

John started to improve at 7 years old. He showed communicative intent, followed instructions, and responded well to the Picture Exchange Communication System (PECS).[5] He engaged in table work, learnt self help skills, performed actions to music, and started using words again.

Discussion

Findings from the clinical assessment showed that John demonstrated relatively typical development for a child with DS until 4 years of age. He showed evidence of regression in development in the areas of communication, social engagement and constructive/imaginative play after 4 years of age. He met the ADI-R and ADOS criteria for autism. On the ABC, John's mother indicated that he displayed high frequency of behaviours in the Lethargy and Stereotypy subscales, which are suggestive of a DS and an autism spectrum disorder (ASD) profile.[6] Collectively, these findings highlight the occurrence of autism-like behaviours in individuals with DS in Singapore.

Although ASD has been found to be 10 times more common in individuals with DS than in the general population, the diagnosis of ASD in DS is challenging, especially in those with significant intellectual disability.[7] For instance, social communication impairments, such as lack of imaginative play, may just reflect profound cognitive impairment.[8] However, this is less likely with a lack of

joint attention. There are some profoundly intellectually disabled individuals with DS who have substantially normal social functioning, albeit at a very low developmental level. A substantial number of people with DS may display stereotyped behaviours, especially those functioning in the severe or profound level of intellectual disability.[9] Careful attention to development and social reciprocity criteria allows a distinction between children with ASD and those with DS and stereotypical movement disorders.[10]

An interesting aspect of this case is John's regression in language and social skills. In about one third of children, autism becomes apparent after a period of relatively normal development but subsequently followed by a loss of previously acquired abilities.[10] This 'autistic regression', is characterised by a loss of previously acquired language skills, social interests and other communicative skills, typically prior to the age of 3.[11] Castillo et al[12] reported that the mean age of language loss in children with ASD and DS was 61.8 months compared to 19.7 months for those with ASD only, suggesting that regression in children with ASD and DS occurs, on average, much later than those with ASD only. In the case of John, his parent reported that he regressed at 4 years 1 month, which was in line with Castillo's et al[12] findings. In addition, John's diagnosis of ASD was only confirmed after the age of 10, demonstrating the challenges of diagnosing ASD in individuals with DS.

At the time of the assessment, John's parents were concerned that his current school placement (although in a special needs setting) would not meet his learning needs. Following the diagnosis of ASD, John was then placed in a structured teaching programme specifically for children with ASD.

Early identification of ASD in children increases the success of interventions.[9] Similarly, this also applies to children with DS with comorbid ASD. Inappropriate educational placement for these children can result in unnecessary emotional stress for parents and may obscure the possibility to recognise ASD. It may be important for primary care practitioners to screen for ASD in children with DS. Increasing the awareness of parents and professionals to the comorbid occurrence of ASD and DS would facilitate early identification and appropriate intervention strategies. This case study adds to the limited but growing literature on individuals with DS presenting with autism-like behaviours after late onset regression.

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