

## Case Report

# Childhood disintegrative disorder with seasonal total mutism: A rare clinical presentation

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### Abstract

Childhood disintegrative disorder (CDD) is a rare autistic-like clinical condition with unknown etiology, in that previously acquired age-appropriate language, social and adaptive abilities deteriorate significantly in 2-10-year-old healthy children, although physical and neurological evaluations display no observable abnormality. Our case is a 22-year-old female born of a consanguineous marriage, with the appearance of CDD symptoms in her fifth year of age following normal mental and physical development during her initial four years of life. Without any precipitating factor, she gradually lost her language abilities, social relational skills, affectionate behavior, adaptive capacities, peer play and meaningful interest in her surrounding, friends and family members over a period of 4 years, reaching a plateau in her ninth year of age. The unique special clinical symptom in this case is a seasonal total mutism, which after the beginning of her CDD symptoms is revealing every year covering the spring. As no additional physical or psychological change accompanies her total seasonal speech loss, it cannot be attributed to any mental condition known as having a seasonal pattern. Because in the literature CDD is presented mostly as case reports with lacking of advanced research data, describing any new case is recommended to improve the knowledge about this rare condition, especially if it displays some new unusual signs, not reported till now.

**Key Words:** Childhood disintegrative disorder, mutism, seasonal, speech loss

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### INTRODUCTION

Childhood disintegrative disorder (CDD), also known as Heller's syndrome, is a rare clinical condition,<sup>[1-3]</sup> which was first described by Theodore Heller in

1908.<sup>[4]</sup> It is characterized by a distinct regression of acquired intellectual, language, behavioral, social, relational and adaptive abilities, following a period of at least 2 years of apparently age-appropriate normal development in all aspects of mental functions.<sup>[1-3]</sup>

According to the Diagnostic and Statistical Manual of Mental Disorders Fourth Edition Text Revision (DSM-IV-TR), to be diagnosed as having CDD: (1) there must be an apparently normal development for at least the first 2 years after birth, (2) clinically significant loss of previously acquired skills has to occur in at least two of the following areas: Receptive or expressive language, social skills or adaptive behavior,

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play, motor skills and bowel or bladder control and (3) abnormalities of functioning has to be in at least two of these areas: Qualitative impairment in social interaction, qualitative impairment in communication and restricted, repetitive and stereotyped patterns of behavior, interests and activities.<sup>[5]</sup> Because after being fully established, CDD is highly similar to autism in clinical aspects, the clear initial period of at least 2-year-long normal development is necessary to differentiate them.<sup>[3]</sup> In the majority of cases, the functional impairment is high and relatively permanent and constant for the rest of the life.<sup>[3,6,7]</sup> This dramatic loss of skills causes great concern for the caregivers, making them consult the pediatrician immediately.<sup>[3,8]</sup>

The etiology of CDD is unknown and no specific medical or neurological cause has been found to account for it.<sup>[6]</sup> Epidemiologic data suggest that one out of about 175 children with pervasive developmental disorder (PDD) have CDD.<sup>[8]</sup> CDD is much less common than autism and research data on it is sparse<sup>[4]</sup> thus making the scientific findings about it to be not as advanced as those of autism.<sup>[8,9]</sup>

Interest in CDD has increased markedly in the recent years.<sup>[7]</sup> In the literature, CDD is mostly described in the form of case presentations.<sup>[7]</sup> Considering the rareness of CDD, reporting any case could be informative,<sup>[3,8,9]</sup> especially if it presents with some new, not yet reported clinical symptom in addition to the ordinary signs of CDD.<sup>[8,9]</sup>

## CASE REPORT

This special case of CDD is a 22-year-old female born of a consanguineous marriage (her parents are the cousin of each other) at full-term normal delivery, with birth weight of 4000 g and an Apgar score of nine. There is no history of a pre-, pri- or post-natal complication recorded in her and her mother's health evaluation file, routinely filled in health service centers for any Iranian pregnant woman from gestation until 2 years after delivery and all Iranian children from birth until their high school entrance. She had normal physical and psychological developmental milestones till 5 years of age and her language development, social interactions, communication skills and adaptive behavior were age appropriate until that time.

According to her parents' report, at the age of 5 years, she gradually developed a deteriorative change in her language abilities and social relational skills, without any known precipitating factor. A significant decrease was seen in her relating to people, other children and even her parents and siblings, and her

eye contact and affectionate behavior diminished dramatically. Showing a silly affect, no meaningful interest in the surrounding was exhibited by her, but some echopraxia and frequent affectless staring at people's face. She seemed self-absorbed, not interested in activities she had previously enjoyed (e.g., TV watching, games, peer relation,...) was occupied with her own world, doing some stereotypic behavior like body rocking and self-laughing and some ritualistic, obsessive and compulsive like behaviors in the form of repetitive washing. Her speech was progressively lost and became restricted to echolalia and a few simple, in part unintelligible, words and sentences. Phonological complications worsened the ambiguity of her speech.

This gradual pervasive regression, which continued over the following 4 years, reached a plateau at her ninth year of age. Her adaptive functioning and purposeful activity became limited to handle her self-care and some simple personal and home chores. However, she could eat independently, dress and undress by herself and her elimination skills were preserved. There was no change in her sleep, appetite or other biological cycles and she displayed no signs of hyperactivity, irritability, mood or psychotic change as well as hallucination or delusion. There was no history of abuse (of any type) or any other trauma.

There was no history of any systematic abnormality, prominent infectious disease, serious medical illness, head injury or convulsion and also no positive finding in physical examination, auditory tests, EEG, brain computed tomography, magnetic resonance imaging, routine laboratory tests (kidney function test, liver function test, cell blood count, electrolytes, fasting blood sugar and serum lipids) and metabolic tests. Her body mass index was 23 and she had normal menstrual cycles without any detectable psychiatric change in relation with them.

She had two siblings. There was no history of abnormal neurologic, genetic, medical or psychological conditions in her parents, sister, second generation and extended family, but a 19-year-old brother who is mentally retarded due to unknown origin. Her family dynamics, including the power hierarchy, interpersonal boundaries and relations, member roles and family rules, are in a fairly normal and structurally unremarkable range and she lives in an ordinary and almost open family without significant expressed emotions.

Not being able to study in an ordinary school due to her regression in intellectual, cognitive, lingual, behavioral and adaptive abilities, she is attending an exceptional school till now, in which non-pharmacologic therapeutic and rehabilitative

services specialized for children with mental retardation (MR) and those with PDD were included and provided. In a recent evaluation by the Wechsler test, her IQ was 70 (+/-5).

The unique clinical presentation in this case is her seasonal mutism. We visited her in Rafsanjan city in the fall of 2012. She had displayed a regular, yearly repeating, 3-4 — month long total mutism, from end of winter to beginning of summer, covering the spring, each year since the beginning of her CDD disorder (16 times till now). No precipitating factor could be found for this periodic yearly total speech loss except for seasonality. All medical evaluations were normal at that time (like other times) and her EEG showed no change or finding during that time. No further change could be found in her cognitive, emotional, behavioral, vegetative (sleep pattern, appetite, energy and psychomotor pattern) and menstrual pattern and in her family and living situation during that period.

Antipsychotic medications (several typical and atypical types), mood stabilizer (Valproic acid) and antidepressants (several types of selective serotonin reuptake inhibitors SSRIs and tricyclic antidepressants – TCAs) are frequently tried in low to even moderate doses and for appropriate duration in order to control some of her behaviors, improve some of her abilities and treat the seasonal mutism since the beginning of the CDD symptoms, but could not end up in any change. This was the same as with non-pharmacological interventions. Sensory integration therapy (SIT), behavioral interventions in types of applied behavior analysis (ABA) within the range of her capabilities and behavior modification techniques in order to reduce problem behaviors and prompt adaptive behaviors, social skills training in types of picture exchange communication system (PECS) and functional spontaneous communication skills training, play therapy and floor time (all accompanied with parent education and training) are among the main interventions that are carried out for her, but with rather poor to minor positive response.

## DISCUSSION

The dramatic deterioration of social reciprocity, verbal and non-verbal communication, adaptive and affectionate behavior and progression to autistic-like symptoms after a 5-year-period of normal age-appropriate physical and psychological development, along with normal medical and neurological examination and normal laboratory tests and brain imaging suggest CDD diagnosis in this case.

She is born of a consanguineous marriage. The role of genetic inheritance as a predisposing factor for CDD is under study<sup>[6]</sup>; however, for autism, there is strong evidence that it aggregates in certain families because of shared genes.<sup>[10-12]</sup> The presence of two disordered children (one with CDD and one with MR) in this family may be suggestive of some genetic abnormalities exacerbated due to consanguinity.

The treatment of CDD is similar to autism. Highly structured behavior-based treatment plans including speech, language and occupational therapy, social skills development, educational interventions and parent training are emphasized and are useful to some extent in CDD patients.<sup>[2]</sup> Pharmacotherapy with atypical antipsychotics and SSRIs has shown varying degrees of effectiveness in controlling behavioral problems in PDD patients; therefore, they are used in patients with CDD as well.<sup>[2,13]</sup> However, in this case, none of them (neither pharmacologic nor non-pharmacologic interventions) could cause some fairly positive changes in her clinical condition as it is not uncommon for therapeutic intervention to fail in PDD patient.

The regular, yearly repeating, seasonal mutism covering mainly the spring is the special clinical presentation in this case, which is not reported in any CDD case till now. Researchers are interested in studying seasonal effects on mental disorders.<sup>[14]</sup> Seasonal changes in mood, behavior and vegetative functions are well documented.<sup>[14,15]</sup> The literature suggests a seasonal pattern in some mental disorders, especially affective disorders,<sup>[16,17]</sup> as well as eating disorders,<sup>[18,19]</sup> suicide attempts<sup>[20]</sup> and mental hospital admissions.<sup>[21-26]</sup> Studies show a seasonal variation in melatonin and serotonin, which may be linked to seasonal pattern seen in some mental disorders.<sup>[27]</sup> A spring peak in completed suicide,<sup>[20]</sup> hospital admission of bipolar patients<sup>[21-25,28]</sup> and mood-worsening in some depressed patients is reported in the literature.<sup>[23,26,29]</sup> The effect of the increase in sunlight<sup>[26,29,30]</sup> and environmental temperature<sup>[23,24,26,31]</sup> is hypothesized to be related to psychiatric changes during spring. Mood sensitivity to high pollen count, known as spring's allergen, is another hypothesis associated with seasonality of mood.<sup>[29]</sup> In this case, no additional clinical change could be found in any aspects of her mental or physical condition, except of seasonal mutism. Because signs of those psychiatric conditions that are reported to follow a seasonal pattern in a part of patients are not accompanying mutism in this case during spring, her seasonal total speech loss could not be related to or interpreted by any known medical or psychological condition.

## CONCLUSION

The limited number of CDD case presentations in the literature<sup>[3,9]</sup> makes any new case report important and useful in advancing the knowledge about this rare, not yet fully documented, pervasive developmental disorder.<sup>[3,7]</sup> Seasonal mutism without any accompanying physical and psychological signs and symptoms is not reported in the literature till now. Occurrence of this phenomenon in a CDD case makes the condition more discussable.

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