Case Report

Childhood Disintegrative Disorder: A Case Report

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Childhood Disintegrative Disorder (CDD), a clinical syndrome distinct from childhood autism, is a rare unremittingly pervasive developmental disorder resulting from disintegration of mental functions and progressive neurological abnormality. This rare condition is characterized by regression or loss of previously acquired language and social skills after a period of at least 2 years of normal development. This report presenting a case of a 10-year-old boy who presented with normal development until 3-4 years of age followed by gradually developmental deterioration in previously acquired social skills, language and intellectual functions with aberrant behaviors suggestive of childhood disintegrative disorder. This case is reported as a very rare case and there is no previous official report in Thailand.

Keywords: *Childhood disintegrative disorder, Heller's syndrome, Dementia infantilis,Pervasive developmental disorder*

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Childhood Disintegrative Disorder (CDD) has been recognized for many years, was first described by a Viennese remedial educator Theodore Heller in 1908 under the name of dementia infantilis, 35 years before autism was described by Leo Kanner and Hans Asperger[1,2]. The children he found had insidiously developed a severe regression of mental and adaptive function between the 3rd and 4th years of life after normal development[3-5]. CDD is a complex disorder that affects many different areas of the child's development. It is a rare condition manifesting unremitting progressive neurological disorder in children who have developed normally up to at least two to three years old. The patient usually presents with regression or loss of previously acquired language, social functions, motor skills and cognitive function[6]. The clinical presentations of CDD and autism are strikingly similar but CDD differs from autism in the pattern of onset, course, and outcome. CDD begins later in life than autism, the average age of onset was 3.21 years[6,7]. The regression is more global and more severe and seizures are more frequent than in autism. The prognosis of this condition is usually much worse than for autism[4]. Recently, it was classified as a sub-group of autism spectrum disorders (ASD) or pervasive developmental disorders (PDD) in the fourth edition of the Diagnostic and Statistical Manual of Mental Disorders (DSM-IV)[2,8]. It has long been debated whether it is a discrete disorder or simply a late-onset variant of childhood autism. Recent studies suggest that CDD should be classified as a distinct condition[2,4]. The specific cause of CDD is unknown. However, CDD is usually also associated with seizures or abnormal epileptiform activity on electroencephalography (EEG)[4]. Research findings suggest, however, that it may arise in the neurobiology of the brain[9]. The prevalence of CDD is quite rare; it is 60 times less than that of autistic disorder. The estimate prevalence from four surveys is 1.7 per 100,000. The ratio of boys to girls is estimated to be 8 to 1[10,11]. The following case is a 10-year-old male child diagnosed with CDD.

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Case Report

A 10- year-old Thai boy consulted from Department of Pediatric Neurology due to his aberrant behavior and gradual loss of speech. The patient was born of a full term normal delivery with no perinatal complications and had normal developmental milestones especially acquired age appropriate social and communication skills until the age of three to four years. He was toilet trained and was able to control his bowels and bladder. He also attended kindergarten school wherein he learnt to recite consonants and vowels. He also used to be able to take care of himself. At the age of 3 to 4 years, he gradually stopped performing tasks that he used to be able to such as

asking for snack or taking personal hygiene. His speech also deteriorated to a few unintelligible words. A change in his behavior was noticed when he gradually lessened relating to people as well as to his family members. He would remain occupied in his own world doing some repetitive purposeless activity like continually walking around or grasping stuff. He was most likely to reveal or accentuate his instability, impulsivity and inattention. He sometimes showed emotional dysregulation with irritability and frequent temper tantrums. He even lost the toilet training he acquired previously and started to pass stools and urine in clothes. He stopped attending school and his parents took him to see a Pediatricians. He was diagnosed with autism. He was referred to a higher center wherein he was given a medication (0.5 mg/day of risperidone) and special training for autism. After the treatment, he was somewhat improved.

At the age of 8 years he started to develop the first generalized tonic clonic seizures where were then absent for approximately one year. The patient from then on started to get severe seizure attacks frequently. He was then referred to the pediatric neurologist at Thammasat University Hospital. All the clinical laboratory investigations for the possible causes were done but all came out normal; however, his parents were very burdened by his symptoms and behavior.

On mental state examination, he looked quite a bit younger than his age. His eye contact was poor and he would not maintain eye-to-eye contact. He would continue to go on doing aimless activities without showing any interest in his surroundings. He would engage in some play activities if directed to do so, but would not seek out other children. He was verbal with limited language skills. He communicated mostly by pointing and using one or two word phrases and would only speak to voice demands. He displayed self-stimulatory behavior in the form of walking around and grasping things with his palm. His gross motor skills were below normal and his balance was not as good. He had frequent tantrums and often demanded attention.

The EEG showed slow waves of posterior dominant rhythm, focal slowing and polyspikes at his left hemisphere. The EEG result supports focal epilepsy with mild encephalopathy. An additional MRI study was performed from which no demonstrable abnormalities were detected. The pediatric neurologist started an antiepileptic drug, levetiracetam tab. 500 mg combined with sodium valproate solution (200 mg/ml). Dosages were gradually adjusted to control seizures effectively.

The dosage of resperidone was changed to be 0.5 mg twice a day and over the next 2-3 weeks from which he showed some improvement. The symptoms of anger, irritability and impulsivity diminished. The child was scheduled for clinical assessment once a month with the above medications and specific training to assess the improvement. Until now, 2 years into treatment, follow-up visits have taken place and trivial improvement was found in the above-mentioned areas. Providentially, there have been no seizure attacks for more than 1 year. The care givers' burden has, thus, been relieved by family psycho-education, counseling and supportive psychotherapy.

Discussion

Childhood Disintegrative Disorder (CDD) used to be part of a larger category called autism spectrum disorder[6,7]. It is a neuropsychiatric neuro-developmental disorder. Unlike autism, children with CDD show severe regression after several years of normal development and a more dramatic loss of skills than a child with autism does.

The cause of CDD is still unknown. It is a rare disorder for which not enough research data to determine its cause. The high incidence of epilepsy suggests that the cause of the disorder could be of abnormal neurobiological origin. However, experts do not know whether epilepsy plays a role in or is the result of the disorder[8]. In addition, comprehensive medical and neurological investigations in children diagnosed with CDD hardly uncover an underlying medical or neurological causes. Affected children with CDD are usually brought to see a pediatrician because of the severe loss of their previously acquired social and language skills. After thorough examination and investigation, if there are no organic causes to explain the condition, the patients will be referred to a child psychiatrist.

According to DSM-IV TR[7], to be diagnosed with CDD, a child must show an autistic-like picture and a marked regression or a dramatic loss of age-appropriate normal development for at least the first 2 years. The regression usually occurs in more than two of these areas: 1) communication skills (expressive or receptive language), 2) social skills, 3) play skills, 4) motor skills, or 5) bowel or bladder control. In addition, the patient must show abnormal functioning in at least two of the following areas: 1) Impairment in social interaction (e.g., impairment in non-verbal behaviors,

failure to develop peer relationships, lack of social or emotional reciprocity); 2) Impairment in communication (e.g. delay or lack of spoken language, inability to initiate or sustain a conversation, stereotyped and repetitive use of language, lack of varied make-believe play); or, 3) Restricted repetitive and stereotyped patterns of behavior, interests and activities, including motor stereotypes and mannerisms. None of the impairments should be better explained by schizophrenia or other pervasive developmental disorders. The regression or loss may occur abruptly over the course of days to weeks or gradually over the extended period of time. Developmental screening or tests seem to be the most effective intervention to look for underlying medical or neurological conditions that may be signs or symptoms leading to making accurate diagnoses. Most of the patients were diagnosed as autistic. Detail information of developmental history was crucial in making a precise diagnosis. A comprehensive treatment plan should be provided as early as possible in order to prevent further deterioration of the patient.

There is no cure for CDD. The emphasis falls on early and intense psycho-educational interventions to the parents. Treatment involves a combination of medications, behavior therapy and other approaches such as speech therapy, occupational therapy, social skills development according to the needs of the individual child[10]. Treatment for the disorder is basically to relieve or lessen symptoms and to prevent further deterioration of the patient. The roles of pharmacological interventions such as atypical antipsychotics or selective serotonin reuptake inhibitors can be beneficial for behavioral problems. In this case study, the patient's irritability, impulsivity and emotions were improved after taking resperidone, an atypical antipsychotic that works through the dopamine and serotonin systems. The antiepileptic drugs used in this case (levetiracetam and valproate sodium) also help to prevent him from severe seizure attacks. In contrast, the outcome of the patient's abilities and behaviors were only marginally improved by consistent and combined behavioral interventions from caregiver and health care team members.

Conclusion

CDD is a rare but stern condition. Unlike autism, CDD shows severe regression of acquired skills after several years of normal development. Developmental history and screening is crucial for early detection and awareness of the disorder. Children with CDD generally need lifelong support with the activities of daily living and long-term care facility. Coping and support for the family members is also one of the most imperative issues for the CDD affected family. However, early detection and early comprehensive intervention can help improvement and alleviate deterioration for children with CDD.

What is already known in this study?

The clinical presentations of CDD and autism are strikingly similar but CDD differs from autism in the pattern of onset, course, and outcome. Does it really matter if the child has Autism or CDD? At the individual level, based on current available treatment interventions, the answer is no. The utility of differential diagnosis is more relevant to research, as it defines a possible source of heterogeneity that may inform the researcher on developmental trajectories, causes, and treatments for different types of autism-like childhood conditions.

What this study adds?

This is the first official case report of CDD in Thailand. A limitation of our case report is the detailed information about diagnosis before the patient was referred to our department; however, the information presented by the patient's mother was sufficient for a more precise diagnosis and help to understand the clinical spectrum of this rare disease. Overall, we regard the benefit of the early detection, patient-centered and systems-oriented approach as obvious in our complex case. More research and in-depth evaluation are needed for the specific contribution of the causes and the art in improving healthcare for children with CDD.

Potential conflicts of interest

None.

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Childhood disintegrative disorder: รายงานผู้ป่วย

ไพรัตน์ ฐาปนาเดโชพล

Childhood disintegrative disorder (CDD) เป็นลักษณะของกลุ่มอาการของโรคซึ่งต่างจากโรคออทิสติกในเด็ก เป็นโรคของความผิดปกติด้านพัฒนาการแบบรอบด้าน ชนิดที่พบไม่บ่อยและอาการมักไม่ดีขึ้น ความผิดปกตินี้เป็นผลมาจากการที่สมองไม่สามารถทำงานประสานบูรณาการกันและมีความผิดปกติทางระบบประสาทมากขึ้นเรื่อย ๆ ความผิดปกติที่พบไม่ได้บ่อยนี้ จะมีลักษณะถดถอยหรือสูญเสียด้านภาษาและทักษะทางสังคมที่ได้พัฒนามา ตามปกติแล้วอย่างน้อย 2 ปี รายงานนี้นำเสนอกรณีผู้ป่วยเด็กชายไทย อายุ 10 ปี มาพบแพทย์ด้วยมีพัฒนาการปกติมาได้อายุ 3-4 ปี จากนั้นพบว่าค่อย ๆ มีการเสื่อมถอยของพัฒนาการด้านสังคม ภาษา และสติปัญญาอย่างมาก รวมทั้งยังมีพฤติกรรมที่ผิดปกติไปจากเดิมเข้าได้กับโรค childhood disintegrative disorder ผู้ป่วยรายนี้ถูกจัดว่าเป็นกรณีที่พบไม่บ่อยและไม่เคยปรากฏรายงานอย่างเป็นทางการมาก่อนในประเทศไทย