



## **Case Report: Childhood Disintegrative Disorder in a 17 Year Old Male Nigerian Adolescent**

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### **Authors' contributions**

*This work was carried out in collaboration between all the authors. Author CD designed the study and wrote the protocol. Authors CD and OC managed the literature search. Author CD wrote the first draft of the manuscript with assistance from author OE. All the authors read and approved the final manuscript.*

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**Case Report**

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### **ABSTRACT**

This case report aims to increase the awareness and hence enhance early detection of Heller's syndrome by health professionals.

Childhood disintegrative disorder otherwise referred to as Heller's Syndrome or disintegrative psychosis is a pervasive developmental disorder characterized by marked regression in several areas of functioning after at least 2 years of apparently normal development.

In this study, a case report of a 17 year old male Nigerian adolescent whose mental illness has lasted for about 15 years is presented. Prior to his birth, his mother experienced prolonged and difficult labour. The patient suffered an episode of childhood convulsion. Nevertheless, he developed apparently normally until the age of 2 yrs 2 months when he lost his previously acquired language, couldn't attain any social adaptation, with associated inappropriate response to environment cues. This patient was also mentally retarded.

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Heller's syndrome is a very rare condition globally; it's often confused with Infantile autism and has a worse prognosis than Infantile autism. Its aetiology is unknown but Central Nervous System pathologies as well as autoimmune factors have been implicated. Early detection as well as concerted team effort is needed to improve treatment outcome.

*Keywords: Heller's syndrome; disintegrative psychosis; development; pervasive; disorder.*

## 1. INTRODUCTION

Childhood disintegrative disorder is also known as Heller's syndrome after the Viennese educator, Theodore Heller who first described the condition in 1908 [1]. Heller described six children who had insidiously developed a severe mental regression between the 3<sup>rd</sup> and 4<sup>th</sup> year of life after previously normal development. He therefore called it dementia infantilis. This syndrome also referred to as disintegrative psychosis or social development regression was identified 35 years before Leo Kanner described Autism; but it was not officially recognized until recently [1].

Childhood disintegrative disorder (CDD), Autistic disorder, Rett's syndrome as well as Asperger's syndrome are all referred to as autistic spectrum disorders. These disorders were fused into a single diagnostic term "Autistic spectrum disorder (ASD)" under the DSM V manual on May 15, 2016 [2,3]. The global prevalence of ASD is 1% [4]. These disorders are characterized by abnormalities in communication and social interaction and by restricted repetitive activities and interest [5]. In addition, Heller's syndrome is characterized by marked regression in several areas of functioning (especially language, social play and adaptive behaviour, motor skills as well as bowel and bladder control) after at least 2 years of apparently normal development [2].

Childhood disintegrative disorder is an extremely rare condition globally and published report of this disorder in Nigeria is virtually non-existent. This fact, coupled with the tendency for this very devastating condition to be misdiagnosed because of the similarity of its clinical presentation with that of autism, informed our interest in presenting this case report. This will make for early detection of cases and hence appropriate management. Before the presentation of this case report for publication, informed consent was obtained from the mother of the patient. This is in line with the ethical guidelines of the University of Port Harcourt Teaching Hospital where this case was managed.

## 2. PRESENTATION OF CASE

A 17 year old male Nigerian adolescent presented with a more than 14½ year history of loss of intelligible speech, aggression, restlessness and other irrational behaviour.

Patient's earlier history was within normal limits. Prior to his birth, the mother had an uneventful pregnancy; however he was born by spontaneous vertex delivery after 18 hours of prolonged labour. Developmental milestones were normal and he was fully immunized. In fact, he was reported to have walked at 9 months. By age 2, he was speaking in sentences and his development appeared to be proceeding appropriately. At age 26 months, he was noted to abruptly exhibit a period of marked behavioural regression. He lost his previously acquired skills of communication as well as bladder and bowel control. He responded to questions occasionally with a nod, though nonchalantly and sometimes with monosyllabic unintelligible speech. Patient became uninterested in social interaction. Auto-aggression in the form of biting himself or banging his head against the wall to express his annoyance when provoked, as well as physical aggression towards other people and destruction of property became evident. There were also associated restlessness, laughing without motivation and such stereotypic behaviour as rocking to music, bruxism and wringing his fingers periodically.

Apart from an episode of childhood convulsion at age 5, no other childhood illness was reported. However, academically he did not progress beyond the nursery school.

The patient was the only child of his parents in a monogamous setting. Family history of mental illness was denied but there was a protracted marital disharmony that eventually led to the separation of his parents. However, the temporal relationship between the onset of the marital discord and that of the patient's illness is not clear.

On mental state examination, a young boy, big for his age came into the consulting room in the company of his mother, laughing to self occasionally, restless, exhibiting repetitive stereotypic wringing of his fingers especially those of his (R) upper limb. He answered almost every question with nod or 'fine' nonchalantly.

Physical examination was essentially normal except that the patient was obese (weight=125kg;BMI= 43.3) with raised blood pressure of 140/100mm Hg. Comprehensive medical and neurological examination as well as radiological examination (CT scan) revealed no identifiable pathology.

Before he established contact with us, the patient had been to several hospitals where many drugs including the antipsychotic Olanzapine had been prescribed at various times. The names of most of the drugs as well as the dosage could not be recalled by informants. However he had not been on any medication for about one year prior to the patient's first contact with us, a year ago. Unsatisfactory medical intervention had led the mother to seek for unorthodox solution to the patient's illness (in the form of prayers). This measure did not yield the expected result either. Nevertheless, mother reported that at age 10, there was some improvement in patient's bladder and bowel control and now he appeared less aggressive than before.

On account of abnormalities in communication and social interaction with associated marked regression in several areas of functioning (language, social play, adaptive behaviour as well as bladder control) after age 2yrs 2mths of normal functioning, a diagnosis of ASD (the CDD variety) was made, in line with the DSM V criteria [2,3], by the attending qualified consultant psychiatrist.

Patient was placed on Risperidone, Carbamazepine and Vitamin B complex tablets. Regular supervised physical exercise aimed at weight reduction as well as the antihypertensive Nifedipine were prescribed. Behaviour therapy programmes were also instituted and a special school for the disabled was recommended for him. However, there has been little improvement so far in his clinical state prior to this report.

### 3. DISCUSSION

Childhood disintegrative disorder is a very rare condition globally. It is estimated to be at least

one tenth as common as childhood autism and the prevalence has been estimated to be about 1 to 100,000 children. It occurs more in boys than in girls by a ratio of 4-8:1 [6,7,8]. Various prevalence rates of ASD have been reported; in the USA, 14.6/ 1000 [9], not significantly different from European rates [9], and in Asia, 26.4/1000 [10]. By contrast, in many parts of the world including Africa, prevalence estimates are either unavailable or preliminary [11].

The onset of this patient's condition was at age 2 years 2 months; this was after a period of normal development. This paints a vivid picture of childhood disintegrative disorder; typically, language, interest in the social environment and often toileting and self care abilities are lost. Hence the patient showed similar social and communication deficits as those associated with autism; the basis of the normal antecedent developmental history [2].

The severe speech impairment is a manifestation of a severe cognitive defect. This patient's speech regressed from age appropriate conversational interchange to a repetitive monologue. He manifested auto-aggression and physical aggression to a significant degree. Violent or injurious behaviour such as banging his head against a wall and biting himself when provoked may be as a result of low tolerance for frustration. This is similar to the clinical presentation of autistic children and this finding agrees with the thought of some researchers who summarized it thus, "some of these patients show an aggressive behaviour, especially tendencies such as beating themselves or head banging" [12]. Furthermore, Chukwujekwu D.C. and Stanley P.C. noted that, the severity of psychopathology, the burden of mental illness as well as deterioration of the patient's personality may among other factors be responsible for this [13].

This 17 year old patient manifested such stereotypic behaviour like rocking to music, wringing of his fingers as well as repeated bruxism. This is in consonance with reports of other studies which state that children with Heller's syndrome are more likely than autistic children to show fearfulness and early stereotypical behavior [14].

This patient was obviously mentally retarded as demonstrated by his extremely poor academic performance. This is yet another proof of his severe cognitive impairment, similar to the

presentation in autistic children. The average reported prevalence of learning disabilities among children with ASD across studies is 52.6% [15]. The degree of intellectual impairment in children with childhood disintegrative disorder appears to be more than that of autistic children, although the overall degree of impairment and outcome appears to be similar in both groups [15].

Only one episode of febrile convulsion was recorded for this patient. This is contrary to research findings on Heller's syndrome. It has been documented that epilepsy occurs much more frequently in children with this disorder than in autistic children and the risk of seizure increases throughout childhood, peaking at adolescence [1,16].

The etiology of childhood disintegrative disorder is unknown but several lines of evidence suggest that it arises from some form of central nervous system pathology. Some consider the condition to be childhood dementia, suggesting that brain deposition of amyloid is the cause of the condition although no clear cut pathophysiology is proven [17]. This abnormal deposition of lipid in the brain is a rare group of inherited metabolic disorders. Childhood disintegrative disorder has been associated with sub-acute sclerosing panencephalities; a chronic infection of the brain caused by a form of the measles virus that results in brain inflammation and the death of nerve cells [1,6]. However, this possible etiology is highly unlikely in the present case because the patient was fully immunized. Furthermore, childhood disintegrative disorder has been linked to tuberous sclerosis (TSC) [1,6]. TSC is a genetic disorder in which non-cancerous (benign) tumors grow in the brain and other vital organs. The computerized tomography scanning carried out in this patient revealed no pathology however. Some experts also suspect that an autoimmune response may play a role in the development of childhood disintegrative disorder.

The stereotypic disorders noticed in this patient further reinforce the suspicion of a neuro-pathological basis of this illness. It has been reported that neurobiological factors including excessive dopamine activity may be contributory in the development of stereotypic movement disorder [18]. Dopamine agonists induce or increase stereotypic behaviour whereas dopamine antagonists decrease them. Endogenous opioids also have been implicated in the production of self injurious behaviours [19].

There was some improvement with the introduction of pharmacotherapy as evidenced by slightly better bowel and bladder control than at the onset of the illness. The antipsychotic Olanzapine was replaced with Risperidone on account of the obesity observed. Risperidone prescribed for 9months was used to treat behavioural abnormalities such as restlessness, irritability, aggression and deliberate self harm. This patient was clearly obese and this can be attributed to the long term use of Olanzapine which has the propensity for inducing excessive weight gain which may be difficult to reverse [20]. Risperidone has been documented to be effective in improving behavioural symptoms in patients with pervasive developmental disorders, but there is little evidence of its specific efficacy in Heller's syndrome [7,21].

Patient has been on Carbamazepine for the past one year and it was used here as a mood stabilizer as well as prophylaxis for seizure which is common in children with childhood disintegrative disorder, more so with the positive history of a febrile convulsion in this patient. Carbamazepine was also given to control automatic behaviour such as stereotypic movement disorders as well as episodic aggression. Behaviour therapy programmes were used by the clinical psychologist as well as the caregiver (his mother) to help the child relearn language as well as learn social and self-care skills. These programs use a system of rewards to reinforce desirable behaviour and discourage problem behaviour.

#### 4. CONCLUSION

The sudden loss of previously acquired adaptive behaviour in a child less than five years of age should always raise the index of suspicion of Heller's syndrome as distinct from infantile autism, in the minds of child health care providers. In line with the general maxim in medicine, early detection is key to better outcome of management. Prompt referral to a child psychiatrist is encouraged when CDD is suspected by the attending clinician in general out patient-settings. The outcome for children with Heller's syndrome is usually very poor and even worse than for autistic children [5]. Those with moderate to severe mental retardation or with an inability to communicate tend to have a less favorable outcome than those left with a higher IQ and some verbal communication [17]. However, a concerted, consistent team effort by all health care members may make for better

prognosis than would otherwise have been the case. Furthermore, children with this disorder may need residential care in a group home or long-term care facility.

## COMPETING INTERESTS

Authors have declared that no competing interests exist.

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