Childhood Disintegrative Disorder (Heller's Syndrome)

Synonyms: CDD, dementia infantalis, disintegrative psychosis, social development regression

Definition
Childhood disintegrative disorder (CDD), or Heller's syndrome, is a rare pervasive developmental disorder (PDD) which involves regression of developmental ability in language, social function and motor skills. It is a devastating condition of unknown cause.

PDDs are a spectrum of behavioural problems associated with autism and autism-like syndromes. They include CDD, Rett's Syndrome and pervasive developmental disorder - not otherwise specified (PDD-NOS). CDD is considered a low-functioning form of autistic spectrum disorder. However, autism does not show the severe regression after several years of normal development which characterises CDD, and children with CDD show a more dramatic loss of skills compared with children with autism. CDD also tends to develop later than autism, and can develop very much later (up to the age of 10 years).

Historical definitions
The definition has barely finished evolving, perhaps indicating both the rarity of and the lack of explanation for the condition. From the 1970s the terms 'disintegrative psychosis of childhood' (ICD-9) and 'childhood pervasive developmental disorder (DSM-III) were used. In 1987 DSM-III-R reclassified it as childhood-onset autism. The ICD-10 (World Health Organization 1992) and DSM-IV (American Psychiatric Association 1994) re-named it childhood disintegrative disorder and classified it under the general category of pervasive developmental disorder. Under the proposed DSM-5 revisions, all PDDs, including CDD, will be subsumed under the single diagnostic category of autism spectrum disorders. The rationale for this is the similarity between the disorders, as it is now thought that the symptoms of the disorders are on a continuum with autism.

Pathogenesis
The cause is unknown. CDD occurs in children who have had previously normal development who then appear to regress, sometimes rapidly. The condition can seem to develop in days or develop over time, and most commonly begins in the fourth year of life, although there is some variation. Some consider the condition to be a childhood dementia, suggesting that brain deposition of amyloid is the cause of the condition, but no clear-cut pathophysiology is proven.

Epidemiology
CDD is extremely rare with an incidence of 1.7 in 100,000 children.

Presentation

Symptoms
Affected children show clinically significant losses of earlier acquired skills in at least two of the following:

- Expressive language skills
- Receptive language skills
- Social skills and self-care skills
- Bowel or bladder control
- Play skills
- Motor skills

Abnormal function also occurs in at least two of:

- Social interaction
- Communication
- Repetitive interests or behaviours

The child presents after at least two years of apparently normal development. This occurs usually between the ages of 3 to 4 years, but generally before the age of 10 years.

- The onset may be abrupt or gradual.
- It can be severe enough that children are aware themselves of the regression, and may ask what is happening to them.
- Usually parents and professionals have not previously noticed abnormalities in terms of language and non-verbal communication, social relationships, play, adaptive behaviour or emotional development.
A typical presentation would be of a child who is able to communicate in two- or three-word phrases losing this ability. They would eventually stop talking altogether or retain only fragments of their former speech.

There may be social and emotional problems, such as a child previously happy to be cuddled becoming averse to physical contact.

Some children describe or seem to be reacting to hallucinations.

Comparison with autism

- The patient eventually shows similar social and communication deficits as those associated with severe or Kanner’s autism. However, it is distinguishable from autism on the basis of the normal antecedent developmental history.
- Children with CDD are more likely than autistic children to show fearfulness and early stereotypical behaviours. [6]
- Epilepsy occurs much more frequently in children with CDD compared with autism. [7]
- The degree of intellectual impairment in children with CDD appears to be more 'even' than when compared with autism, although the overall degree of impairment and outcome appears to be similar in both groups.

Signs

- There are no specific confirmatory signs and physical abnormalities are not usually found, although there may be minor abnormalities such as microcephaly or motor inco-ordination.
- Careful CNS examination including fundoscopy is important to detect other possible causes of the symptoms.

Differential diagnosis

The differential diagnosis includes any of the other pervasive developmental disorders (autistic spectrum disorder, Rett's Syndrome, PDD-NOS) or causes of general learning disability. Other specific conditions which need to be ruled out are:

- Mercury poisoning
- Lead poisoning
- Aminoacidurias
- Hypothyroidism
- Brain tumour
- Organophosphate exposure
- Atypical seizure disorder
- HIV infection
- Creutzfeldt-Jacob disease/ new variant CJD
- Other rare metabolic/neurodegenerative conditions - eg, glycogen storage disorders
- Childhood schizophrenia
- Subacute sclerosing panencephalitis (SSPE)
- Tuberous sclerosis

Investigations

Tests to exclude reversible underlying causes of the condition:

- FBC
- U&E/glucose
- LFT
- TFT
- Heavy metal levels
- HIV testing
- Urine screening for aminoaciduria
- Neuroimaging studies

These are normally carried out during initial assessment in secondary care. Electroencephalogram (EEG), MRI or CT scan are likely to be used to ensure an alternative diagnosis has not been missed.

Management

General measures

Therapy is given, as with autism, tailored to the child’s disabilities, needs and educational objectives. This may include:

- Behavioural therapies, such as applied behaviour analysis, which aim to teach the child to relearn language, self-care and social skills systematically.
- Environmental therapies such as sensory enrichment.
- Medication:
  - Risperidone may be effective in improving behavioural symptoms in PDD. [5] However, there is little evidence of specific efficacy in CDD. [6]
  - Other antipsychotics, stimulants and selective serotonin reuptake inhibitors (SSRIs) are sometimes used in expert hands to help in the control of problematic behaviour, particularly aggression. There is a significant risk of neuroleptic malignant syndrome with the use of neuroleptic medication.
  - Epilepsy may require anti-epileptic medication.
**Prognosis**

- Loss of skills often reaches a plateau by around age 10. There may be some, very limited improvement, but this is seen in a minority of cases.
- In the long term, children have similarities to a child with severe (Kanner's) autism with long-term impairment of behavioural and cognitive functioning.
- Effects on intellectual function, self-sufficiency and adaptive skills are profound, with most cases regressing to severe intellectual disability.
- Medical co-morbidities such as epilepsy commonly develop.
- Those with moderate-to-severe mental intellectual disability or with an inability to communicate tend to do worse than those left with a higher IQ and some verbal communication.
- Outlook is poor. Children will require lifelong support.
- Risk of seizures increases throughout childhood, peaking at adolescence, and seizure threshold may be lowered by SSRIs and neuroleptics.
- Life expectancy has previously been reported as normal. However, more recent studies suggest that mortality of people with autistic spectrum disorders is twice that of the general population, mainly due to complications of epilepsy.[6]

**Historical note**

In 1908 a Viennese remedial teacher, Theodor Heller, described six children who had insidiously developed a severe mental regression between the third and fourth years of life after previously normal development. He called it dementia infantilis. Dementia infantilis was first distinguished from infantile autism in 1943, when Leo Kanner postulated that they represented separate diagnoses.

**Further reading & references**


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