VIEWPOINT

In defense of childhood disintegrative disorder

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Diagnostic pattern: Children with Childhood Disintegrative Disorder have severe loss of skills after 2 years of age, leading to symptoms of autism such as repetitive behaviors.

Regression, or the loss of skills, has been associated with autism since **Leopold Kanner** first used the word ‘autistic’ to describe a group of children with abnormal sociability and repetitive patterns of behavior in 1943[1]. Kanner described one of his patients as having “gone backward mentally” for two years. Today, any of the five autism spectrum disorders, as listed in the *Diagnostic and Statistical Manual of Mental Disorders IV, Text Revised* (DSM-IV-TR), may be associated with regression. But one in particular, childhood disintegrative disorder, is defined by it.

A diagnosis of childhood disintegrative disorder requires that a typically developing child over 2 years of age undergo a severe and mostly irreversible regression of developmental gains, including speech, sociability and self-help skills. Theodor Heller first described the disorder as *dementia infantilis* in 1908[2]. Despite significant changes in classification systems since 1908, the clinical features Heller described remain the backbone of today’s definition of childhood disintegrative disorder, which was introduced in the DSM-IV in 1994.

But there is controversy in the autism field about whether childhood disintegrative disorder really is a distinct entity, worthy of its own diagnostic category.

In the fifth edition of the *Diagnostic and Statistical Manual of Mental Disorders* (DSM-5), due to be

published in 2013 and now in draft form, childhood disintegrative disorder will be subsumed into a larger autism spectrum disorders category. The committee responsible for drafting the new criteria writes, “We believe that children meeting the DSM-IV-TR criterion [sic] for childhood disintegrative disorder will now fit well within the new diagnostic criteria for autism spectrum disorders.” The committee explains this further when discussing changes to the autism spectrum disorders criteria in general: “previously, the criteria were equivalent to trying to ‘cleave meatloaf at the joints.’”

Perhaps the existing diagnostic utensils are akin to gilded spoons, able to scoop out a serving of meatloaf but incapable of the precise cuts necessary to address empirical questions. But we believe that the late-onset, well-defined regression found in childhood disintegrative disorder is an example of a well-differentiated phenomenon. Any muddiness about the diagnosis is related to the diagnostic tools rather than the disorder itself.

We believe that childhood disintegrative disorder represents a process sufficient to cause autism, but that it is different from the mechanism that leads to autism in children without regression. As such, it is an important example of the heterogeneity of the autism spectrum. Doing away with a separate diagnosis of childhood disintegrative disorder would hinder research efforts into better understanding autism.

Distinct development:

As the DSM-5 committee recognizes, it has been unclear whether individuals with childhood disintegrative disorder have a late-onset or regressive form of autism, or whether this diagnostic category captures a truly distinct condition that affects certain children.

To examine this, **Fred Volkmar** and **Donald Cohen** compared a group of children with childhood disintegrative disorder, diagnosed using the International Classification of Diseases-10 criteria, to a group of children who were diagnosed with autism after 2 years of age[3]. They found that the children with childhood disintegrative disorder had accumulated more skills than the children with autism prior to regression and had worse speech and intellectual disabilities subsequent to it.

From this, they concluded that childhood disintegrative disorder merits a separate diagnosis from late-onset autism. Subsequent studies supported these findings[4,5,6]. However, there are also children older than 2 years of age who gradually regress, and the rare children younger than 2 years who experience rapid and dramatic regressions like those that occur in childhood disintegrative disorder[7].

Other clinical evidence also supports the distinction between childhood disintegrative disorder and late-onset or regressive autistic disorder. Childhood disintegrative disorder is accompanied by deterioration across multiple domains[8], whereas in autistic disorder, regression is often isolated to language. The onset of childhood disintegrative disorder is rapid in comparison to autistic disorder, which is insidious[3]. Children with childhood disintegrative disorder have seizure disorders and

encephalography, or EEG, abnormalities more frequently, and also have higher levels of anxiety and stereotyped patterns of behaviors[9,10,11].

The fact that these variables are not directly related to the diagnostic criteria for childhood disintegrative disorder supports the clinical validity of the disorder, by suggesting it has distinguishing characteristics in addition to those criteria.

All in the timing:

Psychologist Cheryl Hendry contends, as does the DSM-5 committee, that the diagnostic continuity between autistic disorder and childhood disintegrative disorder undermines the argument that childhood disintegrative disorder and autism are distinct: “Childhood disintegrative disorder should not yet be considered distinct from [autistic disorder], as not enough information exists to justify it as a separate diagnostic category,” she writes in a review published in 2000. Hendry further argues that the diagnosis is an unwarranted division of a cohesive group, pointing to a fundamental problem with categorical diagnostic systems[12].

It is true that to re-conceptualize the autism spectrum disorders along behavioral dimensions, as will happen in the DSM-5, will draw attention to the fact that many of the factors that distinguish childhood disintegrative disorder from autism are differences of degree, rather than kind. These differences of degree might even illustrate the similarities between autism and childhood disintegrative disorder, rather than the differences.

But the unique and dramatic development of the children currently described by childhood disintegrative disorder suggests that there is more to be gained from first recognizing the disorder as a distinct category and then looking at its overlap with other conditions, than from eliminating the diagnosis altogether.

Research conducted by our team at the Yale Child Study Center emphasizes two points relevant to this discussion. First, children with diagnoses of childhood disintegrative disorder undergo severe, rapid regressions. In the group we study, 70 percent have bouts of terror and agitation preceding regression. In our recent translation (in press) of Heller’s 1908 paper, *Über Dementia Infantilis*, we found multiple references to anxiety, terror, motor agitation and generally bizarre behavior, all preceding the loss of adaptive function.

For example, of one of his subjects he wrote: “In the third year of life the child underwent a psychosis. Attacks of severe [night terrors]occurred several times. Later on he seemed to be nervous during the day as well, he talked gibberish, seemed to be afraid and often didn’t recognize his parents.”

Second, children with childhood disintegrative disorder undergo very substantial losses of adaptive function and end up far more disabled than their counterparts with autism and no regression.

The symptoms that precede regression and the devastating nature of the regression itself may reflect the timing of the process: The later you regress, the more you have to lose, and the more dramatic and severe the regression is. After all, we know that the behavioral changes associated with autism are cumulative. For example, cognitive researchers **Warren Jones and Ami Klin** observe that "failure to look at the eyes of others during critical windows of development, and looking at other parts of the world instead, suggests an altered path for learning about the world, with cascading effects on further socialization[13]."

An explanation for childhood disintegrative disorder that depends on an altered path for learning about the world and the cascading effects that this has on subsequent development is not easy to reconcile with its precipitous onset. It seems likely that although childhood disintegrative disorder and autism are, eventually, clinically similar, the natural history of childhood disintegrative disorder marks a unique mechanism, the elucidation of which could inform us about autism in general.

Because of this possibility, childhood disintegrative disorder deserves a high degree of research scrutiny. Without a distinct diagnostic category, clinicians would be less likely to bring unusual, late-regressing children to the attention of researchers. The science will suffer as a result.

From this point of view, not enough information exists to take the risk of eliminating childhood disintegrative disorder as a separate category in the DSM-5. We hope that a more complete understanding of the biological basis of childhood disintegrative disorder will eventually lead to its removal from the DSM-5, as with Rett syndrome, but removing the disorder now will hinder efforts to discover the pathophysiology of this unique and devastating neurodevelopmental disorder.

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