A Case Study of Childhood Disintegrative Disorder Using Systematic Analysis of Family Home Movies

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Abstract Childhood disintegrative disorder (CDD) is a rare pervasive developmental disorder that involves regression after a period of at least 2 years of typical development. This case study presents data from family home movies, coded by reliable raters using an objective coding system, to examine the trajectory of development in one child with a reported regression at 48 months of age. Coding substantiated parent reports of mostly typical early development, followed by later catastrophic loss of skills across many developmental domains. Differential diagnosis of CDD and autism with regression is discussed.

Keywords Childhood disintegrative disorder · Autism · Regression

Introduction

Childhood disintegrative disorder (CDD) is a rare pervasive developmental disorder estimated to affect 1.7 in 100,000 children (Fombonne 2002). It was first described a century ago by Theodor Heller, an Austrian educator, who reported on six children who lost previously acquired skills after a prolonged period of normal development (Heller 1908). The differential diagnosis of CDD is determined largely by its pattern of onset. There must be at least 2 years of typical development (mean 3.4 years; Volkmar 1992), followed by a loss of skills across multiple domains of development, including language, social, cognitive, adaptive, play, motor, or self-help skills (American Psychiatric Association 2000). Autism may also have onset after a period of mostly typical development, but the timing of regression is earlier (mean 18–20 months; Lingam et al. 2003) and many children show subtle signs of delays prior to the regression (Ozonoff et al. 2005; Werner et al. 2005).

In CDD, after the developmental losses, there is typically no further regression (e.g., the condition is not progressive or degenerative), but recovery of function may be limited. After regression and symptom onset, CDD presents very much like Autistic Disorder. Onset may be insidious or abrupt (American Psychiatric Association 2000). Some reports suggest that there is a premonitory phase preceding the regression, characterized by agitation and anxiety (American Psychiatric Association 2000). No clear biological causes of CDD have yet been discovered (Volkmar 1992; Volkmar et al. 2005). There have been reports of psychosocial stressors preceding onset (Kurita et al. 2004; Volkmar 1992), but their etiological significance is unclear.

Several case studies of CDD have been published, with most collecting information through parental report (Bray et al. 2002; Kurita 1988; Russo et al. 1996). In one case, review of home movies confirmed typical developmental status prior to the onset of regression (Volkmar et al. 2005). In this paper, we present a case study of a boy with CDD. Unique to this study, family home movies were systematically analyzed by coders unaware of the child’s diagnosis or the purposes of the study, trained to reliability on an objective coding system (Werner and Dawson 2005). The rates of key social-communicative behaviors are thus
objectively quantified from birth through the period of regression, as described below, validating the clinical descriptions of the syndrome that preceded this report.

Methods

Video was collected as part of a larger IRB-approved study using home movies to study regression (Ozonoff et al. 2008). After obtaining informed consent, families were asked to provide all videotape footage of their child from birth through the time of regression. Footage was transferred from existing formats to DVD and the original media returned to families. Home video footage was cataloged by date, segment start and end time, and number of persons in the frame. A new segment was defined when the events, location, or date of the activity on the video changed. Any segments that did not contain the subject, were undated, or were of poor quality were omitted from further analysis. For the case described here, home movies from 2 to 60 months of age, as well as a 10-min lab-based free-play session recorded at 69 months, were coded (615 min) using Noldus: The Observer 5.0 software. Behaviors coded were gaze to people, non-word vocalizations, one-word and multi-word verbalizations, orienting to name, pointing, repetitive motor movements, repetitive actions on objects, and unusual visual behaviors, using a coding system based on Dawson and colleagues (Werner et al. 2005). Coders were undergraduate students with limited previous exposure to pervasive developmental disorders. They were unaware of the purpose of the coding, the subject’s diagnosis, or the study hypotheses. Coders were trained to identify the presence, frequency, and duration of the coded behaviors using segments from children excluded from the larger study. They were required to establish reliabilities of 80% agreement or higher with standardized training tapes prior to coding participant data. After training, to maintain ongoing reliability, 25% of the data files were double-coded. All Kappa’s were above 0.75 (mean = 0.92).

The participant’s data was divided into six development periods, each ~12 months long, with the fourth period ending around the time the regression began (48 months). To account for differences in the amount of codable footage available in each period, raw duration and frequency scores were converted to proportion and rate scores, respectively. Means for each period were calculated and graphs of the developmental trajectory of five representative coded behaviors are presented in Fig. 1. In the next section, we describe the behaviors coded from home movies, as well as retrospective parent reports from the Autism Diagnostic Interview-Revised (ADI-R), a detailed developmental history, and a comprehensive medical record review. Pseudonyms are used and identifying characteristics disguised to maintain confidentiality.

Case Description

Nicholas was born in May 1998 at 36 and a half weeks gestation. His mother began prenatal care in the second month of pregnancy. There were no complications during pregnancy, labor, or delivery other than supplemental oxygen for 2 days after birth. His medical history for the first 4 years of life revealed nothing significant. In the extended family there is a history of speech/language difficulties but no reports of pervasive developmental disorders. Nicholas’ parents and brother developed typically, with no learning or behavior problems.

0–12 Months

Analysis of family movies found high rates of social gaze and orientation to name during this period. Nicholas was easily engaged, looked at the faces of others frequently, directed many smiles to people, vocalized communicatively, and used toys and objects in a manner appropriate to his developmental level. By parent report, there were no developmental concerns during this period.

13–24 Months

On home video, Nicholas was responsive to social initiations, showed clear interest in people, directed frequent smiles to others, imitated movements such as clapping and dancing, and directed other people’s attention with frequent points and showing. He used a variety of conventional gestures such as waving and blowing a kiss. His first words appeared on video at 12 months and his first independent steps at 14 months. By parent report, he used over 100 words by 2 years of age. Parents reported having mild concerns about Nicholas’ rate of speech development during this period, but after discussing it with their pediatrician, it was mutually decided that no action was needed.

25–36 Months

On video, Nicholas frequently made direct eye contact, shared affect, and communicated, both to request and to direct other’s attention. He used a variety of gestures

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1 The following modifications were made to the coding system of Werner et al. (2005): (1) only gaze to faces was coded from their “gaze at people” code, (2) only pointing was coded from their “gesture/joint attention” code, and (3) their “repetitive motor with and without objects” code was expanded to include unusual visual inspection of objects.
coordinated with gaze. He can be seen pretending and playing appropriately with toys. Language use has increased and Nicholas can combine words into phrases (e.g., “open it” and “a branch and a ball”) and use pronouns. Occasionally, when excited, he is observed on video to jump and flap his hands. While his parents had few concerns at the time about Nicholas, they stated on the ADI-R (conducted at age 69 months) that his eye contact before age 3 was “never that great.”

37–47 Months

On home video during this period, Nicholas made direct eye contact, initiated interactions, communicated fluently both gesturally and verbally, initiated and responded to joint attention, and demonstrated complex social behaviors like showing off. He spoke in full sentences. There are also several instances of Nicholas jumping and flapping when excited.

By parent report, Nicholas experienced a complicated period between 38 and 42 months of age. He went to a camp where he had difficulty relating with older children and was reportedly bullied. Shortly afterward, the family moved to a different state. Also, at this time his mother was in the middle of a difficult second pregnancy. His family describes this period as very stressful for Nicholas, to the point that he displayed behaviors never seen before. He slept less, was very active, was slower to follow directions, needed to be called several times to get his attention, and smeared feces on a few occasions. His parents took him to a pediatric ophthalmologist at 38 months of age; the reasons for referral stated in the report were “recently noted decreased eye contact, possible exophthalmos, and light sensitivity.” Results of this examination were in the normal range.

Despite the environmental stressors and parent concerns, Nicholas looked very social and communicative on home video in the period between 38 and 42 months, frequently directing smiles and speech to others and making direct eye contact. In one video segment, for example, he can be observed pretending he is playing a hockey game, smiling, and looking at other people while saying “Yeah, I won!” and raising his arms in a victorious gesture after scoring a pretend goal.

Nicholas’ parents reported that after the family settled in their new home, he adapted well and returned to his typical behavior. They reported regular use of complex sentences at this age on the ADI-R (e.g., “Grandma gave me an olive and I think there was a pit in it”). Nicholas was toilet trained with full control of bladder and bowel functions at age 3½. No further disruptions were reported when a baby brother was born when Nicholas was 44 months old. In
video taken at this age, Nicholas can be seen affectionately cuddling the new baby, responding to questions, and using a “shhh” gesture paired with eye contact and smiles.

48–60 Months

In May 2002, just days before his fourth birthday, the staff at Nicholas’ preschool suggested that he should be evaluated for emerging behavior problems, including aggression and withdrawal from peers. His parents were not seeing such behaviors at home, but had noticed periods of intense anxiety and panic-like agitation for a couple weeks. Nicholas would stare at himself in the mirror and scream “Bad Nicholas! You are stupid, I hate you” then become agitated and run around the room. He pinched himself and banged his head on walls and mirrors. His parents reported that, although Nicholas had been eagerly anticipating his fourth birthday party, he became highly upset that morning, crying to his parents, “I can not have my party, no, no, I do not want to have it.”

Nicholas was observed at preschool at 48 months of age by a marriage and family therapist, who noted in her report “continuous hand-mouth stimulation…frequently looking sideways out of the corner of his eyes…repeating the last few words spoken to him,” and general lack of response to adults and peers. The examiner felt that Nicholas was capable of “quite sophisticated language and vocabulary,” giving several examples of spontaneous sentences (“I’m sorry I’m hitting you” and “I like Buzz Lightyear, I do not like school”) but noted that language was rarely used to communicate. Shortly thereafter, Nicholas’ parents noticed the same behaviors at home.

Within 2 months, Nicholas’ parents reported that he lost the use of almost all language other than negative self-talk, became totally withdrawn, did not want to be touched, and was no longer toilet-trained. He lost motor skills as well, including the ability to ride a bicycle and eat neatly with silverware. He became clumsy, tripping and dropping things. All imaginative play ceased. Nicholas no longer used favorite toys as they were intended, but instead lined them up and looked at them from peripheral vision.

Home video taken during the period of initial regression was sparse. A 3-min segment from August 2002, when Nicholas was 51 months old, showed him singing to himself and flipping repetitively through a book while pacing around the room. He echoed several questions posed to him. There is little gaze to people or the camera, but Nicholas did respond after his name was called multiple times, making direct eye contact with his father and smiling. He also answered a direct question (“Who’s that?”) with one word (“Woody”).

Figure 1 plots the coded home video data from birth through 60 months (as well as a lab visit at 69 months). It reveals clearly how Nicholas’ behavior changed in his fifth year of life and validates objectively his parents’ report of a regression around the fourth birthday. Dramatic drops in the rate of social gaze, language use, response to name, and pointing are evident, as is a huge increase in repetitive behaviors, especially the visual inspection of objects from the side and very close to the eyes. On home movies taken between 52 and 60 months, Nicholas rarely looked at faces (on several occasions his mother had to turn his head to get eye contact) and did not orient to his name. In family gatherings, he tended to be alone, watching television very close to the screen while jumping and flapping. His play was manipulative and repetitive, primarily looking at objects closely while talking to himself. He frequently held his arms out in stiff postures, made unusual hand movements that he examined from peripheral vision, and paced or wandered around rooms aimlessly.

Nicholas was evaluated at a university medical center at 4 years 3 months of age. The abrupt change in functioning at 48 months was described carefully in the report, but since his parents had noted poor eye contact prior to 3 years, fulfilling the DSM-IV onset criterion for autism, the ultimate diagnoses were Autistic Disorder and Anxiety Disorder NOS. Intensive applied behavior analysis services began shortly thereafter. Nicholas underwent extensive medical evaluation beginning at 49 months of age. Audiometry, neurological exam, sleep and 24 hour EEG, head MRI, high-resolution chromosome and Fragile X studies, and amino acid and organic acid tests were all in the normal range.

61–94 Months

Little improvement was reported by parents in this period. At 5 years 1 month of age, Nicholas was re-evaluated at the same medical center as initially diagnosed. The report concluded that “given [Nicholas’] regression at a late age and continued plateau in functioning, with questionable continued mild loss of skills even with intensive treatment, a diagnosis of CDD may be most appropriate for him.” He was referred to a neurologist for a second opinion regarding diagnosis. Medical work-up was again within normal ranges on all tests. The neurologist concurred with the initial diagnosis of autism, stating “it does appear that [Nicholas] had PDD symptoms by the second year of life. There was concern about his language development, he had poor eye contact and often ignored when his name was being called.” However, a referral was made to the last author’s study of regression. It was at this time that all video of Nicholas from birth through 60 months (reviewed above) was coded, blind to diagnosis, to provide an additional opinion on the timing of the decline and the most appropriate diagnosis.
At 5 years, 9 months (time 1) and 7 years, 10 months (time 2) Nicholas was evaluated as a research participant using the Mullen Scales of Early Learning, the Vineland Adaptive Behavior Scales, the ADOS, and the ADI-R. He showed clear signs of autism during both evaluations. At time 1, eye contact was very limited and Nicholas rarely attended or responded to the examiner. He spoke little, primarily immediate and delayed echolalia, and did not use any gestures. He used objects in repetitive ways, often inspecting them from peripheral vision, and displayed very high rates of stereotyped motor behaviors such as jumping and flapping, mouthing, clapping, hand clapping, and finger flicking. Functional language declined somewhat from time 1, when there were occasional spontaneous functional utterances (e.g., “want balloon”), to time 2, when language was rare and echolalic. Otherwise the time 2 ADOS was similar in both quality and total score to time 1.

Nicholas’ developmental performance at both time points was significantly delayed. On the Mullen, his age equivalent scores at time 1 ranged from 18 months (Expressive Language) to 25 months (Receptive Language). At time 2, age equivalents ranged from 15 (Receptive Language) to 25 months (Visual Reception). Little developmental progress was seen over the 2-year period, with Mullen raw scores remaining largely the same. One exception was a significant loss of receptive language skills between times 1 and 2, with raw scores decreasing from 25 to 16 passes.

Based on the apparent normalcy of social-communicative development in the first 4 years of life and the dramatic loss of skills at 48 months of age, both quantified through careful coding of home video, Nicholas’ diagnosis was changed from Autistic Disorder to CDD at 5 years 9 months of age.

**Discussion**

The present case study allows us to objectively support the onset pattern that has been described in the literature as defining CDD. There was a period of at least 2 years of typical development followed by a period of agitation and intense anxiety and shortly afterward by a marked loss of social, communication, play, and adaptive skills. Despite extensive work-up, no medical causes or correlates have been found.

The regression was quite abrupt in its onset: no substantive concerns were raised until 48 months, but by 50 months, Nicholas was extremely withdrawn, engaged in high rates of stereotyped behavior, and presented like a classically autistic child. Losses continued for at least a year afterward, perhaps longer. Immediately after the initial regression, Nicholas retained some spontaneous language and could use sentences, whereas a year later he was functionally non-verbal. Our testing found a decline in receptive language skills over a 2-year period, with significant drops in raw scores, although most other skills were stable. Improvement has been very limited.

The differential diagnosis of autism and CDD (Malhotra and Gupta 2002; Volkmar and Rutter 1995) was central to this case and, for all children, rests on the accurate identification of the timing of the losses. In retrospect, Nicholas’ parents questioned whether he might have demonstrated subtle differences in gaze behavior before the regression, although at the time concerns were not pronounced. These retrospective questions about his development may have been a natural reaction to the search for answers that can occur when a child develops a devastating illness. Bray et al. (2002) suggested that parents and clinicians may (unknowingly) fit a child’s behavior to an autism pattern, due to the familiarity of this condition. This is understandable, given the low prevalence of CDD: in the vast majority of cases, developmental regression will indeed be indicative of autism, not CDD. However, in Nicholas’ case, home video clearly told a different story and did not validate the retrospective suspicions of early developmental delays. There was no empirical evidence in home movies of limited eye contact in the second or third years of life, for example. It is possible that selective videotaping or averaging of behavior rates across 1-year periods may have masked subtle atypicalities in development that were later retrospectively reported by parents. Nevertheless, the data depicted in Fig. 1 objectively confirm that dramatic atypicalities in development were not present until later, around the beginning of the fifth year of life, substantiating the ultimate diagnosis of CDD.

The difficulty of the differential diagnosis between CDD and autism in Nicholas’ case underscores the need to clarify the onset pattern of these two conditions. In the DSM-IV-TR, there is a 1-year period of overlap for the timing of the regression, such that a child experiencing developmental losses between ages 2 and 3 could meet criteria for either Autistic Disorder or CDD (American Psychiatric Association 2000). Another point of potential overlap is that some reported cases of CDD present with atypical development prior to the regression (Volkmar 1992; Volkmar and Rutter 1995), just as do some children with autistic regression (Ozonoff et al. 2005). Future studies on onset patterns may provide an empirical basis for how to make the criteria for

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2 The Mullen was administered, although it was not appropriate for Nicholas’ chronological age, because he was unable to complete the routing items of the Stanford-Binet and other intellectual tests were not appropriate for his functioning level. For this reason, Mullen standard scores could not be calculated and only age equivalents are reported.
these two conditions mutually exclusive and increase the reliability of this differential diagnosis.

After Nicholas’ diagnosis was changed to CDD, his parents reported that their school district questioned whether services for children with autism would still be helpful. Ultimately Nicholas retained these interventions and is still served as a child on the autism spectrum, but this required explicit intercession by the professionals who care for him. Thus, while we believe that accurate diagnosis is essential for future research, particularly the etiology of CDD, it is also critical that the phenomenological similarities and treatment needs of the two conditions be clearly stated. In essence, after the regression occurs, CDD presents exactly like severe autism and should be treated in the same way.

Review of home video is not a practical method of differential diagnosis in clinical evaluation, but in this case study, objective coding of family movies was critical to accurate diagnosis. This paper demonstrates the validity of the CDD diagnosis, as first described 100 years ago, and helps put to rest any questions about whether CDD exists.

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