Normal IQ in a 55-year-old with newly diagnosed rhombencephalosynapsis

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Abstract

Rhombencephalosynapsis (RS) is a rare congenital disorder characterized principally by agenesis/hypogenesis of the cerebellar vermis and fusion of the dentate nuclei and cerebellar hemispheres. Fusion of the peduncles and colliculi is common, and associated anomalies of the cerebral hemispheres also can be present. Only about 50 cases with RS have been described and the majority of these have been children. While the literature suggests that RS often is associated with behavioral and/or intellectual impairment, no previous report has described overall neuropsychological functioning. This report describes an employed male who was diagnosed with RS by MRI at age 55. The neurological examination revealed only subtle sensory-motor abnormalities and the results of the neuropsychological evaluation were generally within normal limits, with the exception of poor immediate visual memory and motor dexterity. These findings suggest RS is not inevitably associated with substantial cognitive impairment.

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Rhombencephalosynapsis (RS) is a rare congenital disorder of unknown etiology that was categorized as a focal dysplasia in a recent classification scheme for cerebellar malformations (Patel & Barkovich, 2002). Sener (2000) reported that the frequency of RS was 0.13% in a series of 3000 pediatric patients who underwent head MRI, which offers improved visualization.
of posterior fossa structures compared to CT (Patel & Barkovich, 2002; Savolaine, Fadell, & Patel, 1991). The essential anatomical features of RS are agenesis/hypogenesis of the cerebellar vermis and fusion of the cerebellar hemispheres and dentate nuclei. Fusion of the peduncles, colliculi, and thalami and ventriculomegaly are common. Associated anomalies of the cerebral hemispheres have been observed (Boltenstern, Konrad, Uder, & Kujat, 1995; Demaerel et al., 1995; Montull, Mercader, Peri, Martinez Ferri, & Bonaventura, 2000; Scroop, Sage, & Voyvodic, 2000), as have facial and non-cranial abnormalities (Aydinoglu, Cila, & Aktan, 1997; Michaud, Mizrahi, & Urich, 1982; Sener, 2000; Truwit, Barkovich, Shanahan, & Maroldo, 1991). Some patients die early in life due to co-existing anomalies or illnesses (Savolaine et al., 1991; Schachenmyr & Friede, 1982; Yachnis, 2002).

It is only within the past 15 years, with the advent of MRI, that the diagnosis of RS has been made during life (Truwit et al., 1991). The literature suggests RS never has been diagnosed beyond the fourth decade, either by MRI or at autopsy (Guyot, Kuzmierczak, & Michael, 2000; Montull et al., 2000; Scroop et al., 2000). Between Obersteiner's original WWI era description of RS (Verri, Uggetti, Vallero, Ceroni, & Federico, 2000) and the advent of MRI, only about one dozen cases were reported (Michaud et al., 1982; Truwit et al., 1991). Only approximately 35 cases diagnosed by means of MRI have been described (Auber & Barkovich, 1995; Aydingoz et al., 1997; Boltenstern et al., 1995; Demaerel et al., 1995; Guyot et al., 2000; Romanengo, Tortori-Donati, & Di Rocco, 1997; Savolaine et al., 1991; Schachenmyr & Friede, 1982; Scroop et al., 2000; Simmons, Damianio, & Truwit, 1993; Toelle et al., 2002; Truwit et al., 1991; Utsunomiyi et al., 1998; Verri et al., 2000).

It is well known that the cerebellum is essential for the control and integration of movement and that cerebellar disorders often are associated with a motor syndrome that can include ataxia, dysmetria, disordered eye movements, dysarthria, dysphagia and tremor (Schmahmann, 1991, 2004). There is no single clinical syndrome associated with RS, in part because of the range of supratentorial anomalies (Simmons et al., 1993; Toelle et al., 2002; Truwit et al., 1991). But motor impairments of varying severity, alone or in combination, have been observed in many children with RS (Toelle et al., 2002). For example, Demaerel et al. (1995) described two cases, 2 and 6 years old, who presented with feeding difficulties in infancy and ataxia, balance problems, head rolling, and abnormal eye movements later in childhood.

IQ data were provided in three previous case studies of RS (Montull et al., 2000; Savolaine et al., 1991; Verri et al., 2000). In each case, IQ was at the impaired or borderline impaired level. A recent multi-national study described nine children with RS, one of whom had a normal IQ at 3.5 years old (Toelle et al., 2002). That study provided additional evidence that the clinical presentation of patients with RS is variable, but we are not aware of any previous English language report that described an adult RS case with documented normal IQ or any that presented other neuropsychological data.

1. Case report

This study was approved by the Health Sciences Human Subjects Committee of the University of Wisconsin at Madison and this 55-year-old patient provided his informed consent to have his clinical data presented in a medical journal. As a child the patient lived in a foster
home from the time he was 3 years old. He could not provide any information about his biological family. He denied experiencing any trauma associated with his upbringing. The patient did not complete the 8th grade, but later attended vocational school for 2 years. In his early school years, he obtained “fairly poor” grades. He stated that he had difficulty learning to read, has always done very little if any reading, and is a poor speller. He also stated that his mind is “always wandering” and that this has always been the case. He did not attend special education classes. At the time of this evaluation he had been employed for 5 years as a laborer handling machinery. He had previously worked as a laborer in a different field. The patient is divorced, with two adult children, one of whom reportedly has schizophrenia. He reported that in his spare time he watches television and movies, and he has an extensive movie collection.

The patient reported a history of depressed mood. He added that his symptoms became more significant about 8 months prior to this evaluation. He was taking an antidepressant at the time of this assessment. He had participated in counseling, but stopped attending these sessions because of the expense and the fact that he did not consider them to be particularly helpful for his mood. He reported that current alcohol use was only occasional and typically moderate, but he acknowledged excessive alcohol intake in the past. In 1983, he was hospitalized after being found on the floor of a store the day after he had consumed a large quantity of alcohol. No seizure activity was witnessed and a subsequent EEG was within normal limits. The patient had last used cocaine approximately 30 months before this neuropsychological assessment session. His cocaine abuse involved two 6–8 month periods of daily use that bracketed a period of substance abuse treatment. Medical history included pancreatitis.

2. X-ray and CT

The patient underwent a CT scan of the brain in 1983, after the incident described above, which revealed borderline ventriculomegaly. In August 1999, he was referred for a CT scan because of nasal airway obstruction. In addition to a finding of nasal polyposis, the lateral ventricles were mildly enlarged.

CT of the abdomen and spine films were obtained in January 2000 when the patient complained of pain following a motor vehicle accident that did not involve head injury. Abnormalities of kidney structure and vertebral fusion were identified. An additional rib was present on the right side.

3. Neuroradiology

A MRI in March 2002, conducted after the CT investigating airway complaints revealed mild ventriculomegaly, demonstrated multiple intracranial abnormalities. These included complete aplasia of the cerebellar vermis with midline fusion of the cerebellar hemispheres and abnormal orientation of the middle cerebellar peduncles with displacement toward the midline. The collicular plate appeared flattened, with possible fusion of the colliculi. There was moderate ventriculomegaly and apparent absence of the posterior aspect of the septum pellucidum. The hippocampi were described as abnormal and globular. See Figure 1. Incompletely rotated
Fig. 1. Coronal MRI shows complete aplasia of the cerebellar vermis with midline fusion of the cerebellar hemispheres. Abnormalities include moderate ventriculomegaly and apparent absence of the posterior aspect of the septum pellucidum.

hippocampi (Auber & Barkovich, 1995) and underdeveloped temporal lobes (Montull et al., 2000; Truwit et al., 1991) have been described in previous cases with RS.

4. Neurology

The patient complained of a lifelong history of non-progressive gait disturbance and clumsiness. His speech was slightly dysarthric. Coordination was intact, including finger-to-nose testing, heel-to-shin testing, fine finger movements, and rapid alternating movements without evidence of dysdiadochokinesia or dysmetria. Gait was characterized by normal heel and toe walking as well as a good tandem gait, although he did step out once or twice. Toes were equivocal bilaterally. He had decreased distal vibration sense. His mini-mental status examination (MMSE; Folstein, Folstein, & McHugh, 1975) score was 30/30. He was judged to have a non-progressive disorder, with no symptoms related to the ventriculomegaly.

5. Neuropsychology

The patient had no cognitive complaints other than being a poor speller and stated that he had not observed any decline in his abilities. Because of his work schedule, his neuropsychological evaluation was abbreviated. During the testing, he initially appeared mildly anxious,
but seemed gradually to relax. He was pleasant and cooperative and appeared to put forth good effort throughout the testing. He complained of being tired, having come to the evaluation from nightshift work. See Table 1 for tests administered.

The patient’s seven-subtest short form (Pilgrim, Meyers, Bayless, & Whetstone, 2000; Ward, 1990) Wechsler Adult Intelligence Scale—3rd ed. estimated full scale IQ fell within the average range (WAI-III FSIQ = 90). The estimated verbal (VIQ = 94) and performance (PIQ = 86) IQs fell within the average and low average range, respectively. In addition to the seven subtests comprising the short form IQ, the WAIS-III Symbol Search subtest also was administered so that the processing speed index (PSI), based on Symbol Search and Digit Symbol subtest scores, could be determined. His performance on this index fell at the low average level (PSI = 84). The eight WAIS-III subtest scores ranged from the low average (Digit Symbol, Information) to superior (Arithmetic) level. The WRAT-3 (Wilkinson, 1993) reading subtest score was within the low average range (82).

The patient’s Wechsler Memory Scale—3rd edition (WMS-III; Wechsler, 1997b) working memory index (WMI = 108) was within the average range, as were immediate and delayed memory for stories (WMS-III Logical Memory) and a word list (Rey Auditory Verbal Learning Test; Ivnik et al., 1990; Rey, 1964). Immediate recognition memory for faces (WMS-III) was

<table>
<thead>
<tr>
<th>Test</th>
<th>Score</th>
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<tbody>
<tr>
<td>WAIS-III short form FSIQ</td>
<td>95</td>
</tr>
<tr>
<td>WRAT-3 Reading</td>
<td>38</td>
</tr>
<tr>
<td>Boston Naming Test</td>
<td>51</td>
</tr>
<tr>
<td>MAE COWA (C, F, L)</td>
<td>29</td>
</tr>
<tr>
<td>Trail Making Test A</td>
<td>47</td>
</tr>
<tr>
<td>Trail Making Test B</td>
<td>116</td>
</tr>
<tr>
<td>WCST-64 errors</td>
<td>10</td>
</tr>
<tr>
<td>RAVLT: Total Learning</td>
<td>49</td>
</tr>
<tr>
<td>Long-term percent retention</td>
<td>100</td>
</tr>
<tr>
<td>WMS-III: Working Memory</td>
<td>27</td>
</tr>
<tr>
<td>Logical Memory I</td>
<td>41</td>
</tr>
<tr>
<td>Logical Memory % retention</td>
<td>93</td>
</tr>
<tr>
<td>Faces I</td>
<td>22</td>
</tr>
<tr>
<td>Faces % retention</td>
<td>100</td>
</tr>
<tr>
<td>Grooved Pegboard Test</td>
<td>113</td>
</tr>
<tr>
<td>Dominant hand(s)</td>
<td>105</td>
</tr>
<tr>
<td>Non-dominant hand(s)</td>
<td>93</td>
</tr>
</tbody>
</table>

at the impaired level (standard score = 4), but after a 30-min delay he was able to retain in
memory the limited amount of information that he had initially learned.

Even when compared against individuals with a high school education, performance was
within the low average range on the Boston Naming Test (BNT; Goodglass & Kaplan, 1983;
Ivnik, Malec, Smith, Tangalos, & Petersen, 1990) and on measures of visual attention and
visual-motor sequencing (Trail Making Tests A and B; Heaton, Grant, & Matthews, 1991;
Reitan, 1979). Scores on tests measuring executive functioning were at the average level
(Wisconsin Card Sorting Test—64 card short form; Kongs, Thompson, Iverson, & Heaton,
Examination; Benton, Hamsher, & Sivan, 1994). Motor speed/dexterity (Grooved Pegboard
Test; Matthews & Klove, 1964) was at the borderline impaired level for the right hand and at
the mildly impaired level for the non-dominant hand.

6. Discussion

Cerebellar disorders often are associated with disturbances of movement. Many individuals
with RS have motor impairments, albeit of varying range and severity (Toelle et al., 2002).
In recent years it has been hypothesized that there is also a strong cerebellar contribution to
cognition and mood. For example, Schmahmann (2004) suggested that lesions confined to the
cerebellum can be associated with a “dysmetria of thought” that manifests as a “cerebellar
cognitive affective syndrome” consisting of disturbances in executive functioning, spatial cog-
nition, linguistic abilities (see also Cook, Murdoch, Cahill, & Whelan, 2004) and personality.
This cerebellar syndrome is thought to have its basis in disturbance of the corticopontocere-
bellar circuit, through which the cerebellum is connected to autonomic, limbic and associative
regions of the cortex, in addition to sensorimotor cortices (Schmahmann, 1991). The associa-
tion areas that ultimately are connected to the cerebellum via the pons include regions of the
prefrontal cortices, posterior parietal lobes, superior temporal lobe, and parahippocampus.

Offering support for the existence of the cerebellar cognitive affective syndrome, Levisohn,
Cronin-Golomb, and Schmahmann (2000) reported that 32% of a group of 19 children aged
3–16 who had undergone resection of cerebellar tumors, but no irradiation or methotrexate
chemotherapy, demonstrated evidence of this syndrome postoperatively. Their affective
symptoms included irritability, impulsivity, disinhibition and lability. Extensive lesions of the
vermis were associated with the chronic affect regulation changes and older children were
more likely to show behavioral deficits than younger children. The children with postoperative
affective changes all showed declines in language and/or visual–spatial abilities as well.
In addition, Riva and Giorgi (2000) reported that, in a series of children who underwent
 cerebellar tumor surgery, excisions at the vermis were associated with either behavioral
deficits and language disorders, whereas excisions in the cerebellar hemispheres
were associated with lateralized cognitive deficits (e.g., auditory sequential memory and
language processing deficits after right cerebellar surgery and spatial and visual sequential
memory deficits after left cerebellar resections).

Despite his limited education, the patient described in this report demonstrated relatively
mild motor abnormalities and generally intact cognitive functioning. He did score within the
impaired range bilaterally on a test measuring motor speed/dexterity. This deficit appears consistent with the patient’s report of a lifelong gait disturbance and clumsiness and with the cerebellar malformation. His immediate visual memory also was impaired. Although word recognition ability was at the low average level, the score on that test was almost one standard deviation below his short form WAIS-III verbal IQ. In addition, he rarely reads and described himself as very poor at spelling, so it is quite possible that the patient does have a learning disability. It also is possible that his limited reading ability is related to the cerebellar malformation, because a link between dyslexia and the cerebellum has been hypothesized (Nicolson & Fawcett, 1999).

The patient’s history also includes treatment for depressed mood and substance abuse. An extended psychiatric interview was not conducted as part of the neuropsychological examination of our case, but a review of medical records did not reveal any clear-cut evidence of behavioral dysregulation, apart from symptoms related to the depressed mood.

Thus, the cognitive and mood problems present in this case and others with RS conceivably could be considered consistent with the cerebellar cognitive affective syndrome. However, supratentorial anomalies, including malformed hippocampi in this patient, often are associated with RS, and so it is unknown to what degree the cognitive and behavioral deficits might be related to the cerebellar abnormality itself (Savolaine et al., 1991; Simmons et al., 1993).

In the context of the RS literature, this patient’s relatively intact cognitive and motor status is notable and is probably attributable to the fairly limited extent of his extra-cerebellar abnormalities, including limited ventricular enlargement compared to previously reported cases (Isaac & Best, 1987), and adaptability to early cerebellar lesions (Guberman, 1994). Simmons et al. (1993) suggested that the cerebellar hemispheres may compensate for vermian agenesis/hypogenesis, with the degree of compensation being dependent on the integrity of those hemispheres.

The present case is most similar to that of a 39-year-old whose RS was identified after complaints of chronic pain (Guyot et al., 2000). Subtle ataxia of tandem gait was identified. She reported having been clumsy as a child, but was otherwise developmentally normal, with no remarkable medical, family, or social history. Like our patient, this woman had limited supratentorial abnormalities and was employed, suggesting she had average or near-average cognition, but no formal cognitive data were presented. Verri et al. (2000) described a 22-year-old patient who was employed as a laborer, but this patient had borderline impaired IQ and a psychiatric history that included obsessive oral self-mutilation.

In summary, this report is the first to present a RS patient who was diagnosed beyond age forty and has documented normal intelligence as an adult. Consistent with previous findings of grossly intact cognition on the basis of neurological examination in a minority of RS patients, the generally intact performance by this patient on neuropsychological tests provides further evidence that the disorder is not invariably associated with widespread cognitive impairment (Toelle et al., 2002) and can remain undiagnosed well into adulthood.

References


