Case Report

Perceptual learning disability in a case of salt-wasting congenital adrenal hyperplasia

Mona P. Gajre, DCH*, Dinesh Saroj, MBBS, Suchitra Surve, DCH and Mary Kuttikadan, MBBS

Learning Disability Clinic, Division of Neurology and Development, Department of Paediatrics, Lokmanya Tilak Municipal General Hospital, Lokmanya Tilak Municipal Medical College, Mumbai, India

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Abstract

Specific learning disabilities have been rarely reported in congenital adrenal hyperplasia. The impact of hormones on cognition is still not clearly defined, but it is postulated that females with this condition have excess prenatal androgen stimulation, which increases the risk for cognitive impairment. The salt-wasting variety of congenital adrenal hyperplasia is usually associated with a low cognitive profile, and the risk increases exponentially with more episodes of crisis. We report an above-average cognitive profile in a male with salt-wasting congenital adrenal hyperplasia and a perceptual learning disability.

Keywords: Cognition; Congenital adrenal hyperplasia; Learning disability; Perceptual; Salt-wasting

Introduction

Specific learning disabilities are diagnosed in 8–10% of school-aged children. These affect one or more cognitive skills, including problems in reading (dyslexia), motor function (dysgraphia, dyspraxia, developmental coordination disorder), non-verbal learning disabilities (mathematics, written composition, visuospatial skills, socio-emotional ability), executive dysfunction and attention deficit hyperactivity disorder.1

A greater genetic predisposition of males for learning disabilities is postulated to be due to early exposure to androgens in utero.7 Geschwind and Behan’ hypothesized that androgens delay cerebral maturation in the pre- and perinatal periods, especially of the left cerebral hemisphere, which leads to abnormalities in neuronal migration or abnormal connectivity.

The commonest defect in congenital adrenal hyperplasia (90%) is 21-hydroxylase deficiency, which manifests either as a salt-wasting or a simple virilizing form, both of which can cause cognitive impairment.4 The role of hormones in cognition is multifactorial and not clearly defined.
Theoretically, however, in congenital adrenal hyperplasia, excess androgen or the sensitivity of its receptor during gestation delays neuronal maturation. It has been postulated that females with congenital adrenal hyperplasia have excess prenatal androgen stimulation, which increases their risk for cognitive impairment. Children with the salt-wasting type of this condition have been reported to have lower IQs and to have diffuse brain injury due to episodes of hypotension and hyponatraemia, further accentuating their cognitive impairment. Recent studies indicate changes in white matter on magnetic resonance imaging of these patients. In congenital adrenal hyperplasia, both undertreatment (adrenocorticotropic hormone [ACTH] has a role in attention) and overtreatment (glucocorticoids affect memory) result in to cognitive changes and reduced academic functioning.

Case report

A multilingual 14-year-old boy was referred to the learning disability clinic for academic and behavioural issues. 21-Hydroxylase deficiency was diagnosed when he was 1 month old, after a salt-wasting crisis. He had since been on steroids and fludrocortisone but had poor compliance to medications. His hormonal levels at day 40 of life were 17(OH)-progesterone-11, <6 nmol/L; ACTH, 29.8 pmol/L (normal, 2–11); cortisol, 1.18 μg/dL (normal, 5–25); and testosterone, 0.68 nmol/L (normal, <0.3).

At school, he had poor reading fluency, poor legibility and written expression of English but no problems with regional languages. His arithmetic skills were more severely affected, with poor computation strategies, using his fingers to count and difficulty with word problems. He had difficulty in sustaining attention, was easily distracted and had poor impulse control. He had low self-esteem, with a poor body image and low tolerance of frustration. Behavioural problems such as aggressiveness, anxiety and poor social skills were also evident.

On examination, he was obese (98 kg, >90th percentile) although tall (168 cm, >75th percentile), with a body mass index of 33.08. He had Cushingoid facies, hyperpigmented skin, gynaecomastia and abdominal striae, early onset of puberty (8.5 years), a stage IV Tanner sexual maturity rating, excessive facial, axillary and pubic hair, a deepened voice, acne and a testicular volume of 10 ml. Neurological parameters were normal. His recent blood level of 17(OH)-progesterone was 38.6 ng/ml (normal, 0.6–3.4 ng/ml), and serum electrolytes were normal. He had been prescribed both glucocorticoid and mineralocorticoid therapy but had low compliance (last follow-up at 11 years). Informal assessment of reading, writing and mathematics skills raised a suspicion of a learning disability.

As per protocol, the cognitive profile was assessed on the Malin Intelligence Scale for Indian Children. His scores were 111 on the verbal IQ score and 96 on the performance IQ score, indicating a full IQ of 103, i.e. normal intelligence (90–109). The significant 15-point discrepancy between the verbal and performance scores indicates stronger audio-linguistic than visuo-spatial skills. The scores obtained on the tests are shown in Table 1.

Table 1: Quotients for verbal and performance components of the Malin Intelligence Scale for Indian Children.

<table>
<thead>
<tr>
<th>Subtest</th>
<th>Verbal Quotient</th>
<th>Performance Quotient</th>
</tr>
</thead>
<tbody>
<tr>
<td>Information</td>
<td>81</td>
<td>Picture completion</td>
</tr>
<tr>
<td>Comprehension</td>
<td>129</td>
<td>Block design</td>
</tr>
<tr>
<td>Arithmetic</td>
<td>89</td>
<td>Object assembly</td>
</tr>
<tr>
<td>Similarities</td>
<td>119</td>
<td>Coding</td>
</tr>
<tr>
<td>Digit span</td>
<td>138</td>
<td>Mazes</td>
</tr>
<tr>
<td>Verbal IQ</td>
<td>111</td>
<td>Performance IQ</td>
</tr>
<tr>
<td>Full-scale IQ</td>
<td>103</td>
<td></td>
</tr>
</tbody>
</table>

Interpretation of the verbal subtests of the Scale for Indian Children indicated that the boy had average verbal thinking, comprehension and verbal reasoning and a good working memory. The low arithmetic scores suggested that he struggled with numerical reasoning and sequencing operations. His poor scores on the performance subtests indicated poor picture completion and object assembly, implying poor spatial visualization, short-term visual memory, visual perception and visuomotor coordination, which affect speed and accuracy. He scored well in mazes, implying that he performed tasks with planning and foresight.

Visuospatial skills were tested in motor-free tests of visual perception. The boy was below the average age for appropriate standards in visual memory, sequential memory and discrimination. His poor response time suggested a poor visual working memory. Behavioural assessment on the Spence Children’s Anxiety Scale indicated a generalized anxiety state.

Academic achievement was assessed in the revised Woodcock–Johnson psycho-educational battery. The scaled scores for oral language, broad reading, broad mathematics and broad written language were below average, indicating significant potential achievement discrepancy in reading, writing and mathematics. In reading tests, his scores on all sub-tests, such as letter and word identification (for sight word reading), reading vocabulary (for verbal reasoning), passage comprehension (ability to take contextual cues and simultaneously comprehend passage reading) were below average. In mathematics, he had difficulty with percentages, mathematics reasoning and applied problem skills. He had a slow response on all timed tests for reading, writing and mathematics, suggesting processing speed deficits. Additionally, he was anxious during testing, at times distracted and gave up easily, indicating an inability to sustain attention on tasks. Diagnoses of dyslexia, dysgraphia and dyscalculia in a case of congenital adrenal hyperplasia were made, and academic accommodations, such as permission to use a calculator during examinations, were sought.

Discussion

Reading is a complex skill, which involves working memory, attention, phonological awareness and processing speed, and deficits in these attributes leads to reading disorders. Children with working memory deficits struggle with reading, as decoding words requires storage of sounds in short-term memory while subsequent sounds are being decoded. Visuospatial and social executive skills are essential for an individual’s adaptation to the environment, and deficits
severely affect daily functioning. An understanding of spatial relationships is essential for solving arithmetic problems, and affected children struggle with its concept and operations. They have a basic sense of numbers, but slow processing speed leads to poor performance in calculations.\(^1\)\(^2\) The ability to visualize abstract objects in two-dimensional space and to be sensitive to critical differences between them is significantly affected in cases of congenital adrenal hyperplasia.\(^3\)\(^4\)\(^5\) Furthermore, deficits in visuospatial skills hamper the visualization strategy essential in learning, and children with such deficits have difficulty with graphs, charts, diagrams, maps, geometry and algebra. They struggle with games like puzzles, appear clumsy and uncoordinated and thereby have difficulty with social relationships.

Our case was diagnosed as a salt-wasting type of congenital adrenal hyperplasia, with poor compliance with treatment and follow-up. His 17(OH)-progesterone levels were variable and often elevated, indicating under-treatment. Cognitive changes in children with congenital adrenal hyperplasia may be due to either under-treatment or over-treatment.\(^6\) The associated behavioural changes of aggression, anxiety, mood swings and other emotional disturbances might be due to concurrent sexual precocity.\(^7\) Thus, our patient had above-average intelligence and a perceptual learning disability in the academic domains of reading, writing and mathematics. Comorbid conditions such as inattentiveness, generalized anxiety disorder and emotional liability might also have contributed to his poor academic performance.

This case underlines the importance of a holistic approach to the management of congenital adrenal hyperplasia. Early suspicion of cognitive and academic issues should be addressed proactively for optimal educational and psychosocial rehabilitation. Specific recommendations and remediation are essential cornerstones of special educational services.

**Conclusion**

Specific learning disabilities have been rarely reported in congenital adrenal hyperplasia. The salt-wasting variety of congenital adrenal hyperplasia is usually associated with a low cognitive profile. Visuospatial and social executive skills are essential for an individual's adaptation to the environment, and for solving arithmetic problems and affected children struggle with its concept and operations. They struggle with games like puzzles, appear clumsy and uncoordinated and thereby have difficulty with social relationships. Our patient had an above-average intelligence and a basic sense of numbers, but slow processing speed leads to poor performance in calculations. Early suspicion of cognitive and academic issues should be addressed proactively for optimal educational and psychosocial rehabilitation.

**Author contributions**

The concept, drafting and editing of the article and patient care was done by MPG. SS and MK were involved in patient care and preparing of the preliminary draft. DS was involved in preparing the final draft.

**Conflict of interest**

The authors have no conflict of interest to declare.

**References**