PEDIATRIC GRAND ROUNDS

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PGR-1
Megalencephaly-Capillary Malformation Syndrome: Pediatric Neuropsychological Case Report

Objective: Megalencephaly-capillary malformation syndrome (MCAP) is a rare genetic disorder characterized by a spectrum of anomalies including macrocephaly and neurovascular malformations. Symptoms of MCAP include physical abnormalities, neurologic complications, and although developmental delays have been identified; the research is devoid of neuropsychological and neurocognitive data. To date, only 150 cases have been identified. Currently, there is no cure and there remains no clear consensus of the most effective surgical treatment plan. The diverse presentation of children identified proves to be an obstacle when specifying a neurocognitive pattern in MCAP.

Method: The case will review the neuropsychological profile of a 7 year-old girl with MCAP. Neuropsychological evaluation was completed subsequent to diagnosis and following multiple previous neurologically complex surgeries. Testing was complicated by hearing impairment. Parental and teacher reports identified recent cognitive regression in recall of information, word reading, and letter/number sequencing. Results: Neuropsychological findings revealed a WISC-IV GAI at < 1st percentile, a diverse range of scores across the battery, and a splinter skill strength of average to above average on visual memory tests. Performance included below grade level reading and writing, reduced adaptive functioning, and reported executive dysfunction. Neurocognitive deficits revealed diverse, multisystem and multifocal impairments. Neuroimaging will be presented and discussed. Conclusion: Specific cognitive impairments included overall global delay, limited adaptive and academic functioning, as well as executive dysfunction. However, a splinter skill strength in visual memory prompted specific educational recommendations. The neuropsychological evaluation showed significant decline from the previous evaluation and prompted an immediate neurosurgical consult resulting in another corrective surgical procedure.