

Cognition in rare hormonal disorder

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PHP1A (pseudohypoparathyroidism 1A) is a rare genetic disorder that causes short adult stature, multi-hormone resistance, early-onset obesity and cognitive impairment.

Although <u>cognitive impairment</u> is common in patients with PHP1A, there is no information about the severity or specific areas of deficit, limiting the opportunities for appropriate early intervention.

Ashley Shoemaker, MD, MSCI, and colleagues conducted the first systematic evaluation of cognitive function, executive function and adaptive behavior in children with PHP1A. The investigators tested patients with PHP1A, unaffected siblings and controls matched for age, gender, race and body mass index, and they analyzed parent questionnaires.

Patients with PHP1A had lower IQ scores, poorer executive function, delayed adaptive behavior skills and increased behavior problems. They also had increased rates of attention deficit hyperactivity disorder.

The findings were reported in the February *American Journal of Medical Genetics*. The authors recommend that all children with PHP1A be evaluated by early intervention specialists.

More information: Katia M. Perez et al. Cognitive and behavioral phenotype of children with pseudohypoparathyroidism type 1A, *American Journal of Medical Genetics Part A* (2017). DOI: 10.1002/ajmg.a.38534



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