Neuropsychological Profile of a 5-year-old Boy with Gould Syndrome

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OBJECTIVES

Gould syndrome (COL4A1/A2-related disorders) is an extremely rare, genetic disorder characterized by cerebral vasculature defects, ocular dysgenesis, myopathy, and cerebral cortical abnormalities. Many individuals with COL4A1 mutations exhibit seizures, stroke, visual field defects, and hydrocephalus. To date, research regarding neuropsychological sequelae of Gould syndrome has been sparse. Therefore, this presentation aims to present clinical considerations for pediatric patients with Gould syndrome.

RELEVANT BACKGROUND

Patient is a 5-year-old Hispanic boy born at 35-weeks following perinatal stroke, pre-term birth, and suspected cyanosis. After birth, genetic testing revealed a COL4A1p.G1234R gene mutation. Patient's medical history is notable for cerebral palsy, epilepsy, encephalopathy, hypertonia, global developmental delays with language regression, and attention-deficit/hyperactivity disorder. Previous brain MRIs have found decreased white matter density in both cerebral hemispheres. Abbreviated neuropsychological testing was completed during an inpatient pre-surgical epilepsy evaluation. Patient is non-verbal.

BEHAVIORAL OBSERVATIONS

- Watched two cartoons on repeat via iPad during clinical interview with mother
- Made limited eye contact with examiners, but utilized simple gestures
- Became distressed when iPad was taken away or when mother briefly left room
- Required immediate reward following a single task (i.e., one question in a subtest)
- Overtly distracted by external stimuli

NEUROPSYCHOLOGICAL DATA

Primary Test of Nonverbal Intelligence

Index	SS	Age Equivalence
Nonverbal	74	3 years 9 mos

LANGUAGE FUNCTIONING

Receptive One Word Picture Vocabulary Test-4 Spanish-Bilingual Edition

Test	SS	Age Equivalence
Combined	93	4 years 6 mos

VISUAL/MOTOR FUNCTIONING

Beery-Buktenica, 6th Ed

Test	SS
Visual-Motor Integration	45

Lafayette Grooved Pegboard

Trial	SS
Non-Dominant Hand (left)	61

**unable to grasp pegs with dominant (right) hand*

SOCIO-EMOTIONAL FUNCTIONING

BASC-3, Parent Form in Spanish

Clinical Scales	T-Sc
Hyperactivity	51
Aggression	47
Anxiety	39
Depression	44
Somatization	46
Attention	59
Atypicality	45
Withdrawal	50

Validity Index	
F Index	WNL
Response Pattern	WNL
Consistency	WNL

Adaptive Scales	T-Sc
Adaptability	46
Social Skills	27
Communication	32
Daily Living	31

Autism Spectrum Rating Scales, Spanish Form

Composites	T-Sc
Social Commun	64
Unusual Behav	58
Total Score	63
DSM-5 Scales	66

Treatment Scales	T-Sc
Peer Socialization	52
Adult Socialization	61
Reciprocity	68
Stereotypy	59
Behavioral Rigidity	62
Sensory Sensitivity	48
Attention	58

ADAPTIVE FUNCTIONING

ABAS-3, Parent Form in Spanish

Adaptive Domains	SS
General Adaptive	59
Conceptual	58
Social	77
Practical	71

Adaptive Skills	ScS
Communication	1
Funct Academics	2
Self-Direction	3
Leisure	6
Social	5
Community Use	3
Home Living	8
Health & Safety	3
Self-Care	4
Motor	7

INTERPRETATION

Non-verbal intellectual ability was Below Average. Bilingual receptive language was Average. Further, bilateral fine-motor speed and dexterity and visual motor integration were both Exceptionally Low. Standardized socio-emotional questionnaires revealed ongoing concerns related to attention, hyperactivity, and poor adaptive functioning. Diagnostic impressions included Global Developmental Delay and Attention-Deficit Hyperactivity Disorder, Combined Type.

CONCLUSION

Gould syndrome can include a variety of neurological sequelae depending on neurological defects and incidents. These findings are consistent with the "early vulnerability" position of brain development, which proposes that earlier age at brain insult may result in greater impairments in cognitive functioning (Jacomb et al., 2018). Therefore, ***early intervention and targeted long-term interventions*** **appear imperative** to improve functional outcomes for pediatric patients with Gould syndrome.

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