The Importance of Genetic Counseling in Special Schools for Managing Neurodevelopmental Disorders

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Introduction

Genetic Counselors are increasingly integral to teams assisting families facing neurodevelopmental disorder (NDD) diagnoses. They help families understand NDD implications, educate about potential etiologies and genetics, and participate in diagnostic evaluations. Their role includes aiding families in making genetic-related decisions and adapting to disabilities or genetic conditions. Genetic mutations cause 62% of severe intellectual disability cases, and around 40% of Autism Spectrum Disorder (ASD) cases involve single-gene conditions, genetic syndromes, chromosomal abnormalities, or mitochondrial disruptions.

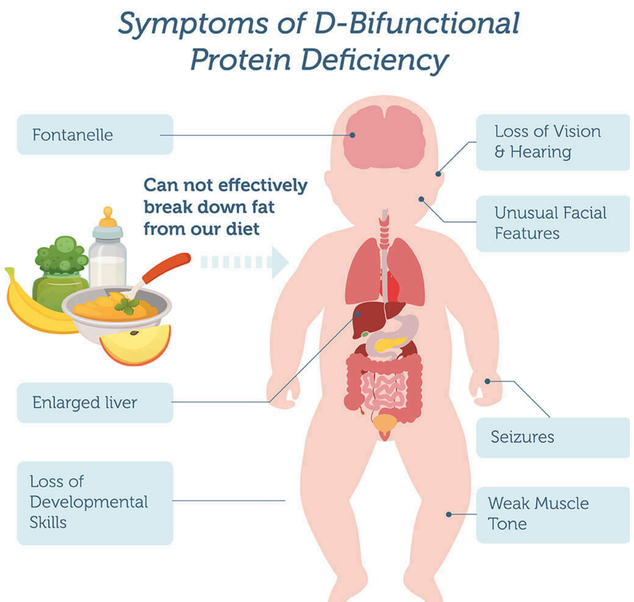
Case Study

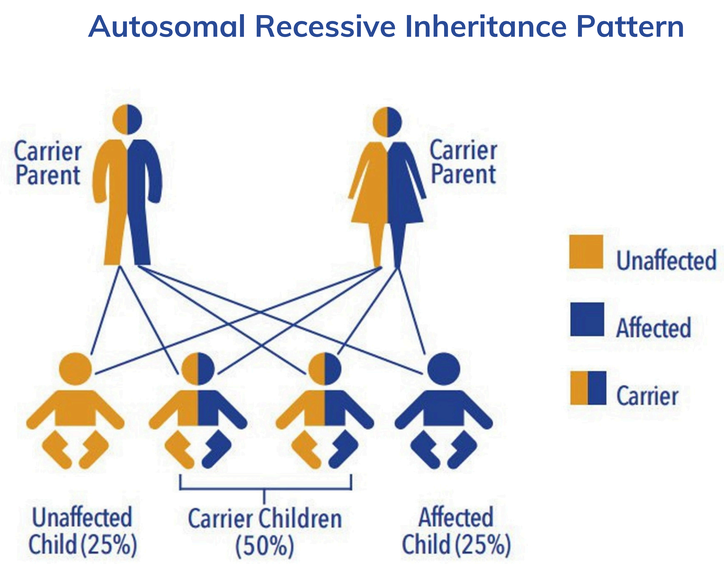
The proband, a 10 year old female child was born to third-degree consanguineous parents, presented with global developmental delay, ataxia, and spastic tone. She had a history of chickenpox, high-grade fever, and seizures at age six, and recent febrile seizures. MRI and MRA of the brain indicated hypoplastic inferior vermis with prominent minor fissures and vallecula, without cystic dilation of the fourth ventricle. Diagnosed with epilepsy with milestone regression, optic atrophy, sensorineural hearing loss and vision loss. She was suspected to have epileptic encephalopathy.

Results

Whole exome sequencing revealed compound heterozygous variants c.43A>G and c.1730T>A of uncertain significance in the *HSD17B4* gene, associated with D-bifunctional protein deficiency. Treatment is supportive, aimed at improving nutrition and growth, controlling the central nervous system symptoms, and limiting the eventual progression of liver disease.

Gene	Region	Variant*	Allele Status	Disease	Classification*	Inheritance pattern
HSD17B4 (+)	Exon 1	c.43A>G **(p.Thr15Ala)**	Heterozygous	D-bifunctional protein deficiency **(OMIM#261515)** Perrault syndrome 1 **(OMIM#233400)**	Uncertain Significance	Autosomal Recessive
	Exon 20	c.1730T>A **(p.Met577Lys)**				





Conclusion

Genetic Counseling and testing in special education settings can aid in diagnosis, prognosis, treatment, and management of NDDs. Identifying genetic etiologies can direct management, offer therapies, and open clinical research opportunities. It can also connect families with support systems and improve the quality of care for affected individuals and caregivers. The clinical features of genetic conditions have multifaceted impacts on individuals' lives, necessitating various forms of assistance and modifications. Genetic evaluations in pediatric populations help identify underlying causes of physical symptoms or neurodevelopmental delays, guiding individualized education programs and specialized care in school settings

Personalized Genetic Counseling, Testing and Care

Reference

Ferdinandusse S, et al Mutational spectrum of D-bifunctional protein deficiency and structure-based genotype-phenotype analysis.PMID: 1638545