

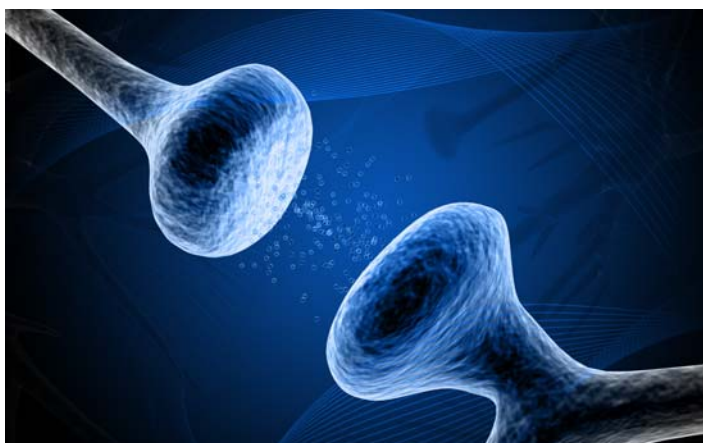
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City of Hope

NATIONAL MEDICAL CENTER AND
BECKMAN RESEARCH INSTITUTE

Novel Methods for Diagnosing and Treating Autism



DESCRIPTION

Autism is a complex neurological disorder characterized by deficits in social interaction, behavior and communication and generally presents in the first few years of life. Autism affects at least 1-2 per 1000 births and its incidence is believed to be increasing. Diagnosis of autism is primarily based on methods assessing behavior in young patients. The **highlighted technology** enhances a physician's ability to diagnose autism by providing a reliable genetic marker that can be assayed easily in a laboratory. The technology adds value by allowing for early diagnosis and; consequently, earlier

intervention and therapy. The technology is based on genetic mutations present in patients with autism. The mutations have been isolated to the Neurexin family of proteins, which acts as pre-synaptic cell surface receptors on neurons and plays an important role in synaptic formation. In a case-controlled study, several Neurexin-1 β missense mutations were shown to be significantly correlated with autism. The incorporation of these missense mutations in an assay could lead to more proficient diagnostics as well as an enhanced understanding of the pathology of autism.

KEY ASPECTS

- Novel missense mutations strongly correlated with autism.
- Mutations present in the Neurexin-1 β gene, which is known to affect synaptic formation.
- Minimizes variability in diagnosis of autism by providing a laboratory-based assay.

INTELLECTUAL PROPERTY

Title	Patent Number	Country	Date Issued
High Frequency of Neurexin 1Beta Signal Peptide Structural Variants in Patients with Autism	7,871,820	US	01/18/2011

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