

SYNGAP1 Dysfunctions as Causative of Mental Retardation

Reference: VAL-656-HSJ

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Background

Mental retardation is the most prevalent severe handicap of children, affecting 1 to 3% of the population. Most patients have the non-syndromic form of the disorder, which is characterized by the absence of associated morphologic, radiologic and metabolic features.

The genetic factors involved in non-syndromic mental retardation (NSMR) remain poorly understood. Linkage and cytogenetic analyses have led to the identification of 29 X-linked and 5 autosomal recessive genes associated with non-syndromic mental retardation, which together account for less than 10% of cases. The present invention offers tools to detect and diagnose further genetic causes associated with NSMR.

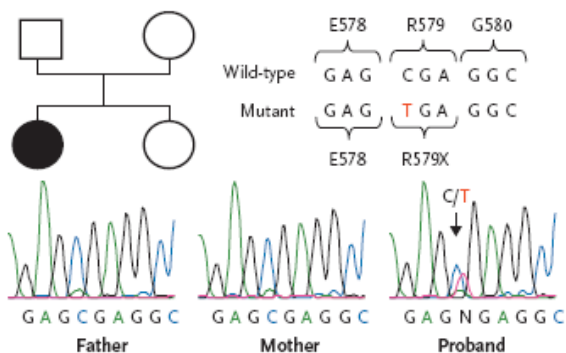
Technology

Dr Jacques Michaud and colleagues from *Hôpital Sainte-Justine* in Montreal-Canada have identified protein-truncating *de novo* mutations in the autosomal gene *SYNGAP1* in 3% of a series of patients with NSMR. *SYNGAP1* encodes a ras GTPase-activating protein that is critical for cognition and synapse function. Besides, extensive screening of patient without NSMR (controls, autism, schizophrenia) did not identify any *de novo* or truncating alteration in *SYNGAP1* reinforcing the premise that disruption of this gene is specifically associated with NSMR.

Results

➤ Proof of concept - Clinical

Clinical Study - Sequencing of the autosomal gene *SYNGAP1* in 94 patients with NSMR identified *de novo* truncating mutations (K138X, R579X, and L813RfsX22) in three of these patients. In contrast, no mutation was observed in 142 subjects with autism disorder, 143 subjects with schizophrenia and 190 control subjects.



Applications

- Diagnostic of non-syndromic mental retardation
- Drug screening
- Therapeutic treatment

Competitive Advantages

- New gene specifically associated with NSMR
- Improved understanding of NSMR
- Better management of patient

Patent Status

PCT application filed (Q4/2009)

Business Opportunity

Diagnostic kit and services for NSMR.

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