Peripheral Biomarkers For The Assessment Of Autism

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SUMMARY

Researchers in the Department of Neurology and the Autism Center in the Semel Institute at UCLA have identified genetic factors which are associated with autism.

BACKGROUND

Autism is a genetic disorder the etiology of which has yet to be clearly described. Recent evidence suggests that the genetic determinants do not follow simple Mendelian inheritance. Instead, the phenotype of autism appears governed by multiple genetic aberrations which together can variably contribute to the severity of the phenotype. Therefore, the identification and examination of these aberrations at both the genetic and functional level will facilitate in the diagnosis and possible treatment of this disorder.

INNOVATION

Researchers in the Department of Neurology and the Autism Center in the Semel Institute at UCLA have identified genetic factors which are associated with autism. Researchers performed genome-wide expression profiling of lymphoblasts from individuals who had either the fragile X mutation (FMR) or 15q11-13 duplication (d15q) and the diagnosis of autism. Dysregulation of two genes were found in common between the FMR and d15q mutations, and were verified in vitro and in vivo models of autism. Furthermore, these two markers were shown to be dysregulated in males diagnosed with idiopathic autism (without known mutations such as FMR or d15q) relative to their non-affected siblings.Further validation of the markers are planned in the near future utilizing phenotype and genotype resources of the Autism Genetic Resource Exchange.

APPLICATIONS

- Examination of these biomarkers will assist in the identification of children at risk for ASD
- Correlative analysis of ASD and these markers may allow for indication of future treatment
- These biomarkers may help identify pathophysiologic targets for treatment development.

RELATED MATERIALS

- Genome-wide expression profiling of lymphoblastoid cell lines distinguishes different forms of autism and reveals shared pathways.

[more]

PATENT STATUS

Country	Type	Number	Dated	Case
United States Of America	Issued Patent	8,173,369	05/08/2012	2007-713

ADDITIONAL TECHNOLOGIES BY THESE INVENTORS

- A Novel Mouse Model for Friedreich's Ataxia
- Biomarkers in Friedreich's Ataxia

INVENTORS

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OTHER INFORMATION

KEYWORDS

autism, genetic diagnostics, biomarkers

CATEGORIZED AS

- **Medical**
  - Disease: Central Nervous System
  - Disease: Genetic Diseases and Dysmorphic Syndromes
  - Research Tools
- **Research Tools**
  - Bioinformatics

RELATED CASES

2007-713-0