

Methods for Determining Autism Spectrum Disorder

Background

Autism spectrum disorder includes autism, Asperger's syndrome and Pervasive Developmental Delay (PDD). Autism is characterized by verbal and non-verbal communication and social interaction deficits and repetitive behaviors. PDD is characterized by sociability and communication delays. Asperger's syndrome is a severe form of PDD but without the language and intelligence impairments associated with autism. Despite intense research in the field, there is no known cause for autism, however, abnormalities in brain structure or function is generally attributed to cause autism.

Currently, there are no reliable diagnostic tests for autism. Diagnosis is often based on observation of patient's communication, behavior, and developmental levels. Since there are other disorders that are also characterized by the behavioral and developmental impairments attributed to autism, diagnosis of autism is often based on excluding these disorders. Thus, there is a critical need for the development of reliable and accurate diagnostic tests for autism. Since research to date indicates a possible genetic basis for susceptibility to autism, a genetic marker would be a valuable diagnostic tool to detect individuals at risk for autism. **The present invention relates to the identification of genes for use as diagnostic tools for autism and methods for identifying subjects susceptible to autism.**

Description of the Technology

The homeobox transcription factor, Engrailed2 (En2), was investigated for possible association with autism spectrum disorder. Linkage/disequilibrium tests in En2 in 137 triads of autistic individuals and their parents indicated that two single nucleotide polymorphisms, Intron A/G and Intron C/T, are critical. The A allele of Intron A/G and the C allele of Intron C/T were transmitted preferentially to autistic individuals in a statistically significant manner, indicating that EN2 locus predisposes individuals to autism. Studies conducted in 29 additional families, including affected and unaffected siblings, confirmed that there is a statistically significant association between susceptibility to autism and the presence of these polymorphisms. Thus, A, C haplotype could be used as a diagnostic marker in the identification and screening of individuals at risk for developing autism.

Advantages

- The A/C haplotype can be used to selectively identify only those individuals at risk for autism.
- Enables early diagnosis and prognosis of autism and this can lead to better healthcare outcome
- The diagnostic tests can be used to advise parents of their likelihood of bearing autistic children.

Applications

- Diagnostic tests for the identification individuals that have autism spectrum disorder.
- Screening tests for the identification of individuals at high risk for autism spectrum disorder.

Patent Status

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- PCT Patent application filed. (Application No.: PCT/US04/21301)

Licensing Opportunity

- This technology is available for exclusive license.

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