

Applications

- Autism Spectrum Disorder (ASD)
- Intellectual disability
- ASD associated epilepsy

Advantages

- Early diagnosis
- Easy genetic testing (mouth swab)

Inventors

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Market Need

Autism Spectrum Disorder (ASD) is one of the fastest growing developmental disorders in the US. Estimated 1 in 110 children is diagnosed with autism and as new diagnostic tools are being developed, these numbers keep going up. Current diagnosis consists mostly of autism-specific behavioral evaluation of toddlers and there is no early medical test available that could allow for “at birth” diagnosis.

Technology Summary

Disturbance in the expression of specific genetic pathways is a significant contributor to autism spectrum disorder. One of the involved genes is *MBD5*, which altered expression is associated with intellectual disability, epilepsy, and autistic features and can be found in approximately 1% of individuals with ASD. This technology uses the analysis of *MBD5* gene expression in different cells (e.g. buccal cells from mouth swab) as an early diagnostic indicator of ASD and can be coupled with direct DNA analysis to improve accuracy of diagnosis.

Technology Status

Patent pending: U.S. rights are available.

Talkowski et al. Am J Hum Genet. 2011 Oct 7;89(4):551-63

Chang et al. Eur J Hum Genet. 2012 Apr;20(4):398-403

This technology is available for licensing to industry for further development and commercialization.