

# ASSOCIATION OF GSTM1 WITH AUTISM AND ASSAYS AND METHODS BASED THEREON

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## Technology description

**Invention Summary:**Autism is a pervasive developmental disorder with diagnostic criteria based on abnormal social interactions, language abnormalities, and stereotyped patterns of behavior evident prior to 36 months of age. Despite its lack of Mendelian transmission, autism is highly genetically determined. Although the vast majority of cases are unrelated to known teratogens, phenotypic expression of autism may be affected by the interaction of environmental factors with multiple gene loci. Oxidative stress could interact with common functional polymorphic variants of genes that protect against oxidative stress and could therefore affect brain development during or after gestation. Glutathione (GSH) is the most important endogenous antioxidant and reports have shown low plasma total GSH levels and elevated levels of oxidized GSH in autism. Glutathione-S-transferases (GSTs) are an abundant and important class of antioxidant enzymes that catalyze conjugation of GSH to toxic electrophiles. Since some genetic polymorphisms of GSTs are known to affect enzyme function, it is possible that a functional GST polymorphism could contribute to the pathogenesis of autism. Researchers at Rutgers have determined that there is a heightened risk of autism for glutathione-S-transferase m genotype GSTM1\*0 homozygotes. The genotype is presumably interacting with other genetic and environmental risk factors. The absence of the GSTM1 gene in GSTM1\*0 homozygotes, because the variant allele is a complete gene deletion that lacks function of the GSTM1 and enzyme could lead to failure in individuals with autism to detoxify important compounds, including some that could be agents or products of oxidative stress.

## Application area

Diagnostic method for autism

Monitor for the effect of pollutants on sample populations

## Institution

[Rutgers University](#)

## Inventors

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