

# NEUROFIBROMIN/DOPAMINE SIGNALING AS A BIOMARKER FOR COGNITIVE AND BEHAVIORAL PROBLEMS IN CHILDREN WITH NEUROFIBROMATOSIS TYPE 1 (NF1)

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T-015422

**Background:** Neurofibromatosis type 1 (NF1) is a common neurological condition affecting roughly 1 in every 2,500 individuals. NF1 is associated with a host of clinical presentations, including malignant tumors and cognitive defects ranging from attention deficits, mental retardation, and autism. Therapeutic intervention has been shown to benefit some of the various cognitive defects associated with NF1, driving a desire for earlier detection of mutations in the Nf1 gene to allow for preemptive interventions.

**Technology Description:** Researchers at Washington University have identified a set of biomarkers correlated with the mutation status of the Nf1 gene, and cognitive ability in transgenic mice. A reduction in Nf1 protein expression resulted in cognitive defects similar to those associated with neurofibromatosis type 1. Blood levels of neurofibromin, dopamine, and key proteins involved in dopamine signaling are usable as biomarkers, suggesting the potential use of the same biomarkers for utility in children with NF1 for risk of cognitive impairment.

**Key Advantages:**

- Allows for early detection of mutations associated with cognitive impairment
- Provides the opportunity for early therapeutic intervention
- Biomarker measurements from bloodstream means minimal invasiveness