Neurogenetic Test Report

Patient Name:	**SIMPSON, Bart**	**GeneDx Accession No:**	**MockTest003**
Date of Birth:	**3/5/1986**	**Date Specimen Obtained:**	**6/2/2013**
Specimen Type:	**Blood in EDTA**	**Date Specimen Received:**	**6/3/2013**
Submitters ID No:	**None Provided**	**Date Test(s) Started:**	**7/21/2014**
Ordered By:	**Dr. Otto Octavius**	**Date of Report:**	**7/22/2014**

Test(s) Requested: TSC1 and TSC2 Genes / Sequencing and Deletion/Duplication Analysis / Tuberous Sclerosis Complex

Describes the clinical features that need to be expressed in the patient in order to meet diagnostic criteria for testing.

Test Indications: Epilepsy, hypopigmented macules, and developmental delay.

Result: **POSITIVE**

Gene	cDNA	Variant	Zygosity	Classification
TSC1	c.1997+1 G>A	IVS15+1 G>A	Heterozygous	Disease-causing mutation

Only mutations detected appear here

No other reportable variants were detected by sequencing and deletion/duplication analysis of the TSC1 and TSC2 genes.

Interpretation: The c.1997+1 G>A splice site mutation has been reported previously in an individual with a clinical diagnosis of tuberous sclerosis (Au et al., 2007). This splice mutation alters the canonical donor site in intron 15. It is predicted to cause abnormal gene splicing, either leading to an abnormal message that is subject to nonsensemediated mRNA decay, or to an abnormal protein product if the message is used for protein translation. The presence of c.1997+1 G>A is consistent with the diagnosis of an tuberous sclerosis in this individual.

Expands on the clinical significance of the identified mutations

Recommendation:

1. Mutation-specific testing for the c.1997+1 G>A mutation in the TSC1 gene is available to the parents of this child for an additional charge to determine if the mutation was inherited or arose de novo. If desired, molecular prenatal diagnosis is available to at-risk family members to address the recurrence risk.

2. Genetic counseling is recommended to discuss the implications of this test report, specifically including the risk of recurrence for this family, clinical variability associated with TSC1 mutations, and testing options for other at-risk family members.

Outlines follow-up recommendations specific to the particular test result, such as references to management guidelines, risks to family members or recommendations for genetic counseling