Novel In-Frame Deletion in *GRIA3*: A Case Report

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X-linked intellectual disability (XLID) is a collection of genetically heterogeneous disorders thought to explain approximately 16% of all intellectual disability in males. More than 90 genes on the X chromosome have been implicated in XLID thus far. One such gene, *GRIA3*, encodes glutamate receptor 3, and is located on chromosome Xq25. Although clinical and functional data on *GRIA3* in the literature is limited, several reports have shown mutations in *GRIA3* to be associated with varying degrees of intellectual disability, behavioral problems, autistic features, seizures, poor muscle bulk, short stature, and dysmorphic features. Here, we present a novel *GRIA3* in-frame deletion located in a functional domain of coding exon 13 (c.2167_2175delGCCCGAGTG), which was identified in a male proband with severe intellectual disability, behavior problems, hypothyroidism, and dysmorphic features. The variant was shown to segregate with disease in all male members of this individual's family, allowing the alteration to be reclassified from a variant of uncertain significance to a variant that is likely to be pathogenic. Previous testing in this proband and his family members detected two translocations [t(4q;10p and t(6;9)] that do not specifically segregate with disease in the family. The data presented here not only contributes additional phenotypic information regarding *GRIA3* alterations to the currently limited literature, but also highlights the importance of family studies in variant reclassification efforts.