Bo's Case	NOTES
Phenotype	A laceration on left index finger with prolonged bleeding previous episodes of prolonged bleeding which hadn't "risen to the level of an ER visit but were concerning." Family history: • No "genetic" family history is available as Bo was adopted from China at the age of 3 years old
Preliminary Diagnosis	Hemophilia (sub-type not determined yet)
Genetic Variation(s)	F9 p.Asp110Gly
Laboratory Assertion(s)	pathogenic
Variant Information: • Asserted interpretation listed in **ClinVar**	pathogenic
• HGVS names from **ClinVar**	NG_007994.1(F9): g.15392A>G NP_000124.1(F9): p.Asp110Gly
• Is population data available in **dbSNP**?	rs137852234 Yes! And it is pretty darn rate.
Gene Information in NCBI Gene: • Symbol and Name	F9 & Coagulation factor IX
• Gene Summary	F9 …vitamin K-dependent coagulation factor IX that circulates in the blood as an inactive zymogen….converted to an active form by factor XIa,…activates factor X … through interactions with Ca+2 ions, membrane phospholipids, and factor VIII. Alterations of this gene…cause factor IX deficiency, which is a recessive X-linked disorder…. [provided by RefSeq, Sep 2015]
• Tissue Expression information	Pretty much just expressed in the liver
• Gene Ontology information	Extracellular Blood coagulation Ca+2-binding & endopeptidase
Ultimate Impacted Biomolecule based on: • **GDV** to view the chromosome and gene region	Located in the coding region within exon 4.
• **RefSeqGene Graphics** view of gene region and transcript(s)	Located in the coding region within exon 4.
• **RefSeq Protein Graphics** view of protein and domains	The protein is made, but with a change in amino acid 110 from an acidic Aspartate to a neutral Glycine.
• **CDD or iCn3D** to view a structure*, as needed*	The variant is identified as one of 3 residues annotated as critical for binding to Ca+2. The change from acidic Aspartate residue to neutral Glycine likely prevents its participation.
Proposed Molecular Mechanism of Variant Impact	This changes an acidic residue which is needed for binding a critical calcium ion which is required for F9 function.
How does this relate back to the phenotype (symptoms/clinical features & diagnosis)?	With the loss of one of 3 coordinating residues for a critical calcium ion, the F9 protein is not fully functional and may not effectively activate the next clotting factor. This correlates with a less severe phenotype than other children might experience.