Spring 2019

Von Willebrand Disease: The Viability of a Technology-Based Diagnostic Modality

Theodore Gutches
Longwood University

Follow this and additional works at: https://digitalcommons.longwood.edu/rci_spring
Part of the Biology Commons

Recommended Citation
https://digitalcommons.longwood.edu/rci_spring/47

This Poster is brought to you for free and open access by the Research & Publications at Digital Commons @ Longwood University. It has been accepted for inclusion in Spring Showcase for Research and Creative Inquiry by an authorized administrator of Digital Commons @ Longwood University. For more information, please contact hamiltonma@longwood.edu, alwinehd@longwood.edu.
**Introduction**

- Von Willebrand disease (VWD) is an autosomal inherited bleeding disorder.
- The VWF is a multimeric plasma glycoprotein that binds to platelets in order to deliver factor VIII, another essential blood clotting protein, to sites in which vascular injury has occurred.[2,4]
- Three types include: Type 1 (partial deficiency), Type 2 (qualitative deficiency), and Type 3 (complete deficiency).
- Type 1, the mild case, makes up 65% to 80% of cases; type 2, the moderate and qualitatively abnormal case of VWD, makes up 20% to 35% of cases; with type 3, the severe case, affecting 1 in 1 million people.[5]
- Variations in Type 2 include: defects in multimerization (type 2A), spontaneous platelet binding (type 2B), defects in ligand binding with multimers (type 2M) and defects in factor VIII binding (type 2N).
- Next-Generation Sequencing has shown promise in its diagnostic application but has not yet been implemented for small-scale patient diagnostics.[1]

**Methodology**

**Patient Population and Blood Sampling**
- Willing patients who have previously been diagnosed with VWD
- ~4mL of blood will be used from each patient

**Amplification and Sequencing**
- Oligonucleotides will be used for amplification of exons
- Multiplex PCRs will be used for amplification of the VWF
- Each amplicon will be processed and analyzed
- A MiSeq platform will be used for sequence analysis[1]

**Sequence Analysis**
- NGS will be used for identification of specific variants, further confirmed via Sanger sequencing
- Categorization of the disease according to type will be recorded

**Potential Conclusions**
- NGS could provide viable results that could advocate for a transition towards a clinically applied system of genetic screening for patients with intermediate forms of VWD
- Identifying new variants among the three types of VWD, through the continued use of gene sequencing for the VWF, could broaden the diagnostic spectrum of VWD, to which clinicians refer.

**Potential Pitfalls**
- Providing necessary tools in order to reduce costs of small-scale genetic testing for patients in a clinical setting could present limitations in the effort for implementation for this diagnostic modality
- Further specification for warranting such a diagnostic tool would require additional evaluations

**References**