The Novel Mechanism of Detecting SV in Bioinformatics Approaches for Preeclampsia Disease

Venkatesh A\*, Poornam. S, Sree Vishmaya.V

Finoseq, RUSA Entrepreneur Hub, Madurai Kamaraj University, Palkalai Nagar, Madurai-625021

Email: **info@finoseq.com**

**Abstract**

Preeclampsia is a significant pregnancy condition that can have a wide range of effects on both the mother and the fetus. Since millennia, there has been no known cure or preventative measure for this pregnant illness. 5-8% of pregnancies are affected with preeclampsia, and the incidence is higher in low socioeconomic areas. According to recent studies, preeclampsia is linked to a higher prevalence of diabetes, cardiovascular disease, and a number of other conditions that affect organs that are more susceptible to damage. According to this, preeclampsia not only exposes the mother and fetus while they are pregnant, but also sets the stage for chronic diseases later on in life. Therefore, comprehensive preeclampsia research is urgently needed since India registers much higher incidence of preeclampsia (15-18%). In Biomedical studies, no predictive assays or biomarkers are currently suitable for early diagnosis for better clinical management. There may be possibilities of identifying novel mechanism of protein misfolding and aggregation underlying the onset of preeclampsia. Protein misfolding and aggregation are the hallmark of neurodegenerative diseases such as Alzheimer's disease. The intriguing data on proteins such as transthyretin and amyloid-beta suggest that preeclampsia is also a risk factor for Alzheimer's disease. Moreover, CRISPR studies also important attention with integration of optical genome mapping (OGM). It may be possibility to find the structural variant (SV) that could be useful by identifying DNA alterations. SVs involve at least 50 nucleotides and due to their size and quantity, SVs act as a significant mutational factor that influences how the genome evolves and functions, as well as how germline and somatic diseases develop. It has been great strides when accessing the advent of long-read sequencing technology, in particular, Pacific Biosciences (PacBio) and Oxford Nanopore technologies (ONT), help to increase the detection of SVs. Furthermore, the advanced technologies such as linked reads (e.g., 10x Genomics) and Strand-Seq has also been developed to improve the quality of assemblies and/or SV calling that could bring new insights into Preeclampsia disease studies. Nevertheless, the increased length and the higher error rate of emerging long-read technologies can pose new methodological challenges and brings attention because the technology and methods are still evolving very rapidly, and the lack of standard protocols needs to be validate for disease studies.

**Keywords**

Preeclamsia, Structural Variant, CRISPR, Optical Genome Mapping, Pacbio, Nanopore, 10x Genomics, Protein Misfolding