HUMAN PREGNANCY-SPECIFIC GLYCOPROTEIN (PSG) GENE VARIANTS AND CNVS ARE ASSOCIATED WITH REPRODUCTIVE DISORDERS
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Success implantation and placentation depend on tightly regulated interactions between fetal trophoblasts and maternal decidua. In humans, pregnancy-specific glycoproteins (PSGs) are important trophoblast-derived effectors which can be detected in maternal serum as early as 7 days post implantation. Earlier studies have shown that injections of anti-PSG antibodies into pregnant animals lead to abortion, and that PSGs could regulate trophoblast invasion and vascular remodeling at the feto–placental interface. In humans, low levels of PSGs have been associated with intrauterine growth retardation and pre-eclampsia, whereas a significant enrichment of cases with deletions in the PSG gene locus is found in pre-eclampsia patients. Because a great number of human PSG genes appear to evolve after the separation of humans from other primates, we hypothesized that genetic variation in PSG genes may have been selected in recent human history and provided survival advantage. Consistently, we have recently reported that human PSG gene region exhibits a significantly higher density of single nucleotide polymorphisms (SNPs) and copy number variations (CNVs) as compared to the genome average. Because some of these variants also exhibit significant population differentiation and signs of positive selection, these variations could be related to heightened risk of reproductive orders. To investigate this hypothesis, we analyzed PSG CNVs in normal and preeclamptic pregnant women. The identification of novel PSG CNV–disease relationships can provide not only a better understanding of the pathology of pre-eclampsia but also a novel opportunity to develop personalized diagnosis for preventing pre-eclampsia and related disorders.