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Abstract Topic:- Molecular and cytogenetic diagnostics

Abstract Title:- Systems biology approach: identification of hub genes, signaling pathways, and molecular docking of col1a1 gene in cervical insufficiency

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Aims:-The collagen type I alpha 1 (COL1A1, OMIM #120150) gene, encoding the alpha-1 chain of type I collagen (UniProt #P02452), plays a crucial role in life-homeostasis due to its involvement in collagen synthesis and the formation of the extracellular matrix. It is a promising candidate gene implicated in the pathogenesis of cervical insufficiency (CI), characterized by premature dilation and effacement of the cervix during pregnancy. This study aimed to identify genetic variations within the COL1A1 gene that contribute to the development of CI.

Methods:- Polymerase chain reaction (PCR) and amplicon sequencing were implemented for single nucleotide polymorphisms (SNPs) detection (+1245G/T, SP1 rs1800012), shedding light on the pathogenesis of CI. Computational approaches viz. Protein-protein interaction (PPI) network, gene ontology, and pathway participation were used to identify the central genes and signaling pathways associated with COL1A1 and CI. Using the online Yet Another Scientific Artificial Reality Application (YASARA) software, molecular docking, and molecular dynamic (MD) simulation with the relevant ligands, oxytocin (CID 439302), estradiol (CID 129728744), progesterone (CID 5994) and hydroxyprogesterone (CID 150788) were carried out to control the state of the COL1A1 protein.

Results:- In the current findings, we found a wild-type sequence for targeted SNPs in enrolled proband indicating that the COL1A1 gene is not involved in the current form of CI. It allows further investigation of other closely related genes probed in this study. Bioinformatics interactive analysis demonstrated that the COL1A1 and more than 10 collagen family genes had a strong connection with CI. In order to get towards therapeutics, the results of molecular docking and MD simulation display the Root Mean Square Deviation (RMSD) value and stability graphs over time for various ligands.

Conclusions:- In sum, the findings of this study provide insights into a modus operandi or approach that can be utilized for the diagnosis of the same. The use of comprehensive molecular analysis and computational studies has illuminated the path toward studying other genes that may be candidates for this condition. These findings have implications for understanding the foundational process of the condition and potentially developing screening, diagnostic, and therapeutic interventions.

Keywords:- Cervical insufficiency, COL1A1, SNPs, Molecular docking