World Academy of Science, Engineering and Technology
International Journal of Medical and Health Sciences
Vol:19, No:03, 2025

Design of a Computational Model to Support the Calculation of a Structural Health Index for Bridges

Authors : Oqba Al-Kuran, Reham Albadaineh, Adnan Almallah, Dana Al-Atrash, Duaa Dahbour

Abstract : Polycystic kidney disease (PKD) encompasses a spectrum of inherited disorders characterized by the formation of multiple cysts in the kidneys and other organs, leading to eventual kidney failure. Autosomal recessive polycystic kidney disease (ARPKD) represents a severe form, typically presenting in infancy with significant renal and hepatic involvement. We report a case of ARPKD in a female fetus, who is a part of twins, delivered at 36 weeks' gestation, exhibiting respiratory distress shortly after birth and subsequent demise. Ultrasonography during the mother's 20th week of pregnancy revealed characteristic findings suggestive of ARPKD in one of the twins. A whole exome sequencing revealed a pathogenic variant in the PKHD1 gene, underscoring the genetic basis of ARPKD. Management involves multidisciplinary care, emphasizing supportive therapy and monitoring for complications. Genetic counseling is pivotal, highlighting the risk of disease transmission and the role of prenatal genetic screening for at-risk families. Our case underscores the importance of early diagnosis, comprehensive management, and genetic counseling in ARPKD, particularly within the Jordanian population, and highlights a novel genetic variant contributing to disease manifestation.

Keywords : ARPKD, Potter sequence, whole exome sequencing, genotype-phenotype correlation, variant
Conference Title : ICOG 2025 : International Conference on Obstetrics and Gynaecology
Conference Location : Istanbul, Türkiye
Conference Dates : March 10-11, 2025

Open Science Index, Medical and Health Sciences Vol:19, No:03, 2025 publications.waset.org/abstracts/190333.pdf