gonadoblastoma. Affected females usually have normal external genitalia and gonads and have only the nephrotic disease. Because they do not have all the features of the condition, females are diagnosed as cases of isolated nephrotic syndrome with persistent proteinuria and abnormal KFT.

Genetic / molecular evaluation: A targeted exome panel (clinical) was performed using illumina next generation sequencing platform from EDTA blood sample for patient. The calculated gender as per the first NGS data output was male (46, XY), however to rule out any sample contamination a repeat NGS run was performed using a fresh EDTA blood sample which predicted the same – genotype of male. Q-PCR was run on the same sample to confirm gender before bioinformatics analysis on the data. Indication based analysis identified a heterozygous pathogenic mutation in WT1 gene - c.1447+4C>T with depth of 11X, indicative of WT1 disorders / a rare genetic disorder of Frasier Syndrome.

Pedigree: