Result: Clinical Exome Sequencing deciphers the complex genetic disorder which was otherwise treated as focal segmental glomerulosclerosis (FSGS). The report also indicated that the sex of the tested individual is male as per the genetic information and hence raised questions regarding the sex of the proband. The proband is phenotypically female and was confirmed by the prescribed clinician, family members and the phlebotomist team. As the outcome of the case was sensitive, the reports were disclosed to the clinician and then to the patient. Currently, the patient is under treatment for disease management. WT1 disorder is inherited in autosomal dominant manner, if the parents are not identified with any variants the risk in siblings is greater than normal population because of possibility of germline mosaicism. However, in genetic counselling sessions conducted for this family other siblings are normal and parents are healthy so further evaluation is ceased considering as incidental finding in proband. Karyotype for the patient is recommended as the next step to rule out true genotype.

CONCLUSION:

Phenotypic correlation in genetic disorder is of utmost importance. For this patient when primary diagnosis of kidney disorder was concluded, secondary symptoms like small uterus, steak gonads were checked in but not recorded and co related. In later follow ups after the results clinician reevaluated the symptoms and amenorrhea was disclosed by patient. WT1 related Steroid-resistant nephrotic syndrome (SRNS) required alternate management often by a multidisciplinary team (medical geneticist, endocrinologist, urologist, and psychologist).

External genital evaluation / checkup is scheduled in the next follow up. Physical features like square face, facial hair and long arms were noticed as later on to check the degree of male genotype expression in the female patient. Otherwise the patient exhibits normal voice and other physical features of a female. This indicates how recording symptoms make an overall impact of the correct diagnosis and correct patient management.