CASE 080

CŌRE DIAGNOSTICS™

AUTHORS: **Ashna Narayan, Ruchi Bhajeewala.**

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CASE HISTORY:

The case is of a 28-year-old female, hails from Srinagar-Jammu & Kashmir. The proband has been presented with kidney issues since last one year; creatinine levels are elevated, presence of protein in urine and suspected focal segmental glomerulosclerosis (FSGS), Genetic testing was prescribed to rule out Alport Syndrome and testing of genes related to kidney disorders. No eyes and ear abnormalities. Case is referred by the clinician for genetic evaluation – clinical exome sequencing to confirm diagnosis when they notice poor prognosis in the patient after one year of steroid treatment. There is no significant family history of similar complaints and as per the information provided by the proband and her sister, there's no consanguinity. The ordered test- Clinical Exome Sequencing came out to be positive for heterozygous pathogenic mutation in WT1 gene which is responsible for WT1 related disorders including Frasier Syndrome-a rare disorder defined by pseudo hermaphroditism and progressive glomerulopathy.

TITLE:

Frasier Syndrome – Decoding diagnosis with clinical exome sequencing – a Next Generation Sequencing approach.

INTRODUCTION:

Frasier Syndrome is a rare genetic condition that affects kidneys and genitalia.

Frasier syndrome is characterized by kidney disease that begins in early childhood. Affected individuals have a condition called focal segmental glomerulosclerosis, in which scar tissue forms in some glomeruli, which are the tiny blood vessels in the kidneys that filter waste from blood. In people with Frasier syndrome, this condition often leads to kidney failure by adolescence.

Females with this condition present with normal female external genitalia, streak gonads, and XY karyotype, and frequently develop