

**Genotype-Phenotype Analysis in an Indian Family Affected with  
Li-Fraumeni Syndrome-Role of Genetic Counselling**

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**KEYWORDS** Germline Mutation. Next Generation Sequencing. Sarcoma. TP53

**ABSTRACT** Li-Fraumeni syndrome (LFS) is associated with the high risk of a diverse spectrum of childhood and adult-onset malignancies with a predominance of the soft-tissue sarcomas, osteosarcoma, breast cancer, brain tumor, adrenocortical carcinomas, Wilms tumor, leukaemia and several other LFS-associated cancer types. This paper reports a case of a 43 years male diagnosed with an undifferentiated, high grade sarcoma. Genetic testing by Next Generation Sequencing revealed a heterozygous likely novel pathogenic germline mutation in the TP53 gene (c.323delG; p.Gly108ValfsTer15) in the proband. Post-test genetic counselling referred the family screening and the other eight family members were found to be carrier for the same variant. Thus the researchers have tried to describe the genotype-phenotype correlation for the LFS with the TP53 mutation which may have contributed to the variable phenotypes in the reported family with reduced/ incomplete penetrance. In this paper the researchers have also tried to highlight the cancer genetic counselling to detect an inherited cancer syndrome