

**UKCGG-National Cancer Genetics MDT meeting, 15<sup>th</sup> October 2020**  
**Li-Fraumeni Syndrome and heritable *TP53*-related cancer (h*TP53rc*) syndrome**

Thank you to all who attended and contributed to the first UKCGG-National Cancer Genetics MDT. We hope that you found it valuable.

Helen Hanson presented an overview of LFS/(h*TP53rc*). Educational slides will be made available on the UKCGG website. Four cases relating to LFS/h*TP53rc* were presented by Guys (Louise Izatt), Cambridge (Ellie Davies), St Georges (Helen Hanson) and Manchester (Emma Woodward). Please contact Helen Hanson ([helen.hanson6@nhs.net](mailto:helen.hanson6@nhs.net)) if you have any cases where choriocarcinoma has been diagnosed in a female partner of a *TP53* carrier. Please contact Ellie Davies ([eleonor.davies6@nhs.net](mailto:eleonor.davies6@nhs.net)) if you have any cases of a *TP53* whole gene deletion. Relevant papers are listed below:

**Risk reducing mastectomy for *TP53***

-Risk of Contralateral Breast Cancer in Women with and without Pathogenic Variants in *BRCA1*, *BRCA2*, and *TP53* Genes in Women with Very Early-Onset (<36 Years) Breast Cancer. Hyder *et al.*, *Cancers* 2020 Feb 7;12(2):378. <https://pubmed.ncbi.nlm.nih.gov/32045981/>

-<http://tp53.co.uk/mastectomy-yes-or-no/>

**Choriocarcinoma risk**

-Transmission of a *TP53* germline mutation from unaffected male carrier associated with pediatric glioblastoma in his child and gestational choriocarcinoma in his female partner. Cotter *et al.*, *Cold Spring Harb Mol Case Stud* 2018 Apr 2;4(2) <https://pubmed.ncbi.nlm.nih.gov/29581140/>

-Transmission of germline *TP53* mutations from male carriers to female partners. Patrier-Sallebert S *et al.*, *J Med Genet* 2015.52: 145–146. <https://img.bmj.com/content/jmedgenet/52/3.toc.pdf>

**Psychosocial**

-Waiting and “weighted down”: the challenge of anticipatory loss for individuals and families with Li-Fraumeni Syndrome. Werner-Lin *et al.*, *Fam Cancer* 2020 Jul;19(3):259-268  
<https://pubmed.ncbi.nlm.nih.gov/32222840/>

-Couples coping with screening burden and diagnostic uncertainty in Li-Fraumeni syndrome: Connection versus independence. Young *et al.*, 2020 *J Psychosoc Oncol*. Mar-Apr 2019;37(2):178-193  
<https://pubmed.ncbi.nlm.nih.gov/30591002/>

-A Systematic Review of How Young People Live with Inherited Disease: What Can We Learn for Li-Fraumeni Syndrome? Forbes Shepherd *et al.*, *J Adolesc Young Adult Oncol*. 2018 Oct;7(5):525-545.  
<https://pubmed.ncbi.nlm.nih.gov/30004834/>

**CHIP**

-Mester *et al.*, Apparently Heterozygous *TP53* Pathogenic Variants May Be Blood Limited in Patients Undergoing Hereditary Cancer Panel Testing *J Mol Diagn* 2020 Mar;22(3):396-404  
<https://pubmed.ncbi.nlm.nih.gov/31881331/>

**Date of next meeting: Thursday December 17<sup>th</sup> 12.30-13.45**