UKCGG-National Cancer Genetics MDT meeting, 20th May 2021
Mixed Cases

Thank you to all who attended the mixed case session on May 2021. Cases were presented from

- St Georges- Dr Katie Snape (familial metastatic gut neuroendocrine tumours),
- Birmingham-Dr Kai-Ren Ong (multiple CRC in a patient with Ulcerative colitis),
- Cardiff-Dr Emily Sloper (multiple primary tumours),
- Nottingham -Ms Catherine Lamb (complexities of breast cancer risk assessment in Trans women)
- Leeds – Dr Alison Kraus (nuchal fibroma in childhood) and
- RMH- Dr Terri McVeigh (paediatric embryonal rhabdomyosarcoma and family history of CRC)
- Professor Marc Tischkowitz also presented his recent paper on clinical management for *PALB2* carriers.

Please send any questions, cases or ideas for future meetings to Helen Hanson
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Relevant papers and resources from the meeting are listed below:

PALB2

-Management of individuals with germline variants in PALB2: a clinical practice resource of the American College of Medical Genetics and Genomics (ACMG). ***Tischkowitz et al***
doi: 10.1038/s41436-021-01151-8.

- **FYI-HBOC** (**F**unctional assa**Y** **I**ntegrated for **H**ereditary **B**reast and **O**varian **C**ancer genes)

http://iscva.moffitt.org/fyi-hboc/build/

-**PALB2 interest group** www.PALB2.org

Breast cancer risk in transgender patients

Breast cancer risk and breast screening for trans people: an integration of 3 systematic reviews (nih.gov)

Health considerations for transgender women and remaining unknowns: a narrative review - PubMed (nih.gov)

Breast cancer risk in transgender people receiving hormone treatment: nationwide cohort study in the Netherlands | The BMJ

A European Network for the Investigation of Gender Incongruence: Endocrine Part - PubMed (nih.gov)

Transgender_cross_programme_screening_leaflet(2).pdf (bhbss.nhs.uk)

Nuchal Fibroma/Gardner fibromas associated with APC

Gardner-associated fibromas (GAF) in young patients: a distinct fibrous lesion that identifies unsuspected Gardner syndrome and risk for fibromatosis - PubMed (nih.gov)

Case Report: Gardener-associated fibroma: an unusual cause of upper airway obstruction (nih.gov)

Identification of previously unrecognized FAP in children with Gardner fibroma (nih.gov)

Management of Familial Adenomatous Polyposis in Children and Adolescents: Position Paper From the ESPGHAN Polyposis Working Group - PubMed (nih.gov)

MDT agreed that in children with isolated clinical features that can be associated with a germline *APC* pathogenic variant e.g CHRPE, Gardner fibroma, desmoid fibromatosis, that if *APC* testing was normal and no pathogenic variants identified , colonoscopy examination would not be indicated in the absence of any further *APC*-associated features or family history

Rhabdomyosarcoma

- Rhabdomyosarcoma
    - Primitive paediatric malignant soft tissue sarcoma, originates from mesenchymal cell
    - Subtypes (WHO classification 2013)
        - Embryonal (~60%)
        - Alveolar (~20%)
        - Pleomorphic (~10%)
        - Spindle/Sclerosing (~10%)

Heritable Predisposition to Embryonal RMS	Common Somatic events
• Beckwith Weidemann S. • dsDNA repair defects Bloom (*BLM*) Nijmegen Breakage (*NBN*) Werner (*WRN*) • Congenital Mismatch Repair Deficiency • DICER1 Tumour Predisposition Syndrome (*DICER1*) • Gorlin S. (*PTCH/SUFU*) • Mosaic variegated aneuploidy (BUB1B) • Rasopathies (*PTPN11, SOS1, BRAF*) • Rubenstein Taybi S. (*CREBBP, EP300)*	• LOH for linked loci at 11p15.5 • ***RAS*** variants • *PIK3CA* variants • *ALK* variants • Trisomy 8