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
helen.hanson6@nhs.net

Relevant papers and resources from the meeting are listed below:

**PALB2**


- FYI-HBOC (Functional assay Integrated for Hereditary Breast and Ovarian Cancer 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)


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%)

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