Moving Genomic Testing to the Mainstream in Paediatric Oncology.

Background:
Cancer syndromes are increasingly recognised in paediatric oncology. Older texts considered hereditary cancer syndromes (HCS) rare, limited to highly penetrant, autosomal dominant conditions, such as retinoblastoma. More recent literature estimates 10 – 29% of children with cancer may be affected by a HCS.

In the clinic, screening for a HCS is often poorly done. The use of a standardised template to assess the need for genetic intervention, administered as part of the initial work up, can avoid this important aspect being missed.

Ideally, all children who have germline mutation testing should see a clinical geneticist or genetic counsellor prior to testing. However, in many centres limited clinical genetic resources would lead to unacceptable delays for both paediatric cancer patients and referrals for other conditions.

Objectives:
To create and implement timely HCS screening for all children with cancer. To support consent and testing for possible HCS by medical and specialist nursing paediatric oncology staff upskilled and supported by Clinical Genetics colleagues. To streamline the referral pathway for families that need clinical genetics referral based on history and exam findings - , separating those for urgent referral (diagnosis may impact treatment options) and routine referrals.

Methodology:
For HCS gene testing, produce information leaflets and consent forms for families. Staff involved will be upskilled in the issues surrounding consent for genomic testing. This will be for pro-band (the child with cancer) testing only and any familial (cascade) testing after a positive result will continue to be managed by clinical genetics.

Children where suggested testing is related to other features such as family history, will be referred to clinical genetics for assessment and testing.

Robust and timely pathways for testing at local and international laboratories will be reviewed regularly.

Outcome:
To date we have created a standardised germline genetics assessment template (GGAT) based on a review of the literature. We have prepared patient information and consent forms (PICFs) for cancer specific testing. Paediatric oncologists have been upskilled in g the process of consent for families and a referral pathway for patients who need direct referral to clinical genetics has been created.

Future Directions:
We are in the process of setting up a project to audit the number of families tested and/or referred to clinical genetics and the number of positive germline predispositions diagnosed against historical controls.