UKCGG-National Cancer Genetics MDT meeting: 16th May 2024

Mixed Cases- led by Glasgow

Meeting contacts:

Dr Rosemarie Davidson (rosemarie.davidson@ggc.scot.nhs.uk)

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Thank you to everyone who attended this session. Below is a summary of the presented cases and relevant publications and resources to refer to.

Presented Cases

Case 1	**FAP/PGD/cord blood**	Presented by: Alina Tsimbaliouk GOSH alina.tsimbaliouk@gosh.nhs.uk
	Actions/Outcome: Notes from discussion: Different to scenario like Huntington's Disease, would not be so dogmatic? Some still feel should include child in discussion and not change view, as issue is funding model for PGT. Other options include prenatal test +/- TOP Issue with testing on a cord sample if gene +ve in newborn period, may be too much to take in at the time, so perhaps timing is the key issue. Could test younger but still have discussion with the child / family at 10-12 about planned surveillance. Store DNA sample for discussion in a few months' time. Some have experienced child being scarred by process at age 10-12 with responsibility to make a decision around testing. Others feel child not so involved in the decision at age 10, so harder to deline testing younger. Ethically if one child has FAP and other is prevented from having FAP by PGD this could cause issues with sibling relationship in future. If don't test as child declines at 10-12 would screen clinically. In case of VHL others have offered testing as young as 6/7years old. Notes from chat: Many have offered cord testing at birth in conjunction with a lot of counselling – case by case decision on whether to test or not at birth. Many have not found testing at age 10 is an autonomous decision for child, more of a family discussion. **Outcome: varied practice across the UK but key theme was around timing of test in at risk child. Experience of testing a child at 10 was mixed. Consensus was no ultimate barrier to offer a test at birth but could offer testing when child a little bit older to**	

	minimise potential trauma to parents in the newborn period (age 2/3 mentioned) so that PGD could be explored as an option if couple wished.	
Case 2	**Reduced penetrance BRCA2 variant**	Presented by: Fiona Beecroft Birmingham fiona.beecroft1@nhs.net
	Actions/Outcome: Notes from discussion: Would advocate high risk breast screening, less strength for mastectomy but it is reasonable to offer this (can always be reviewed in conjunction with patient wishes following reduced penetrance discussion) Notes from chat: Helen Hanson: We have suggested management for BRCA1 R1699Q and would follow this for other variants considered reduced penetrance https://www.ukcgg.org/information-education/national-and-international-guidelines/management-of-carriers-of-variants-associated-with-reduced-penetrance/	
Case 3	**LFS/sarcoma family**	Presented by: Andrew Green Children's Health Ireland (CHI) at Crumlin andrew.green@childrenshealthireland.ie
	Actions/Outcome: Notes from discussion: 1 family with clinical LFS but no variant on LFS register – at risk individuals being offered whole surveillance recommendation Notes from chat: You could ask Hannah Titherage at Birmingham if she would consider the case for nanopore study From Joseph Christopher to Everyone: @Andrew Green we have a LFS sequencing project in Cambridge which are recruiting families without molecular confirmation. Happy to discuss further - jc27@sanger.ac.uk. It would also be very useful to do WGS on the relapse lesions from the affected daughter (we would need fresh frozen tissue) but we can arrange this	
Case 4	**TP53 VUS**	Presented by: Claire Peyton Hermitage clinic, Ireland cpeyton@hermitageclinic.ie
	Actions/Outcome: Very similar to previous case in terms of suggestions- re surveillance	
Case 5	**5q31.2 deletions including CTNNA1**	Presented by: Rachel Harrison Nottingham Rachel.Harrison@nuh.nhs.uk
	Actions/Outcome: 5 families presented with similar deletion, possible founder in the region. Concern about diffuse gastric cancer risk but little information on penetrance, so best practice remains uncertain.	

Case 6	**Ferguson-Smith**	Presented by: Claire Searle Nottingham claire.searle@nuh.nhs.uk
	Actions/Outcome: Post meeting suggestion from Zosia Miedzybrodska in Aberdeen: would a path review by specialist pathologists in Dundee be helpful and David Baty re testing? Post meeting email from Terri McVeigh: *"my lady was 73 by time I met her – she had seen Dr Rona MacKie and the Prof Ferguson Smith before I got near her so was quite well informed about the whole thing.* *We discussed Generalized eruptive keratoacanthomas of Grzybowski as an ddx but dismissed it as invasive SCCs are not common.* *She was labelled as dermatitis artifacta as a long time because of the skin picking issue but she has histologically proven keratoacanthomas and SCCs.* *No variants in TGFBR1 identified but they didn't do dosage analysis so that's one thing to consider at least for my case.* *Not sure that helps with your case but they are strikingly similar in appearance."*	
Case 7	**Mosaicism**	Presented by: Terri McVeigh Royal Marsden terri.mcveigh@rmh.nhs.uk
	Actions/Outcome: Cases to be rolled over to next meeting	
Case 8	**[TOPIC]**	Presented by: [name] [organisation] [presenter email address]
	Actions/Outcome:	

Relevant publications/resources

Topic	Link

Expert Speaker Talk

[Emma Cowan- Westray BRCA1 variant

The slides can be accessed at: emma.cowan2@nhs.scot

Talk Summary:

Summary of work going on in Aberdeen – North of Scotland Regional Genetics Service in relation to Orkney Westray BRCA1 variant testing.
Upcoming plans to expand testing to individuals in Scotland with at least one grandparent from Westray, will be doing so region by region.
Aberdeen happy to be contacted if anyone unsure what to do if requested to have testing for this variant.

Meeting Summary

(If relevant)

Next meeting details

Date	18th July 2024
Time	12:30 pm-1:45 pm
Theme	To report or not report: Variants outside the test directory e.g. ATM c.2251-10T>G
Leading centre	Liverpool
Contact for cases	**Rachel.hart@nhs.net**

Please send any questions or ideas for future meetings to Helen Hanson (helen.hanson@stgeorges.nhs.uk)