IACC 2023 Case-based discussion (CBD) scenario

Specialty:	Genomic Counselling

CBD Scenario

CBD Scenario Title	Take informed consent from a patient for whole genome sequencing (WGS)
CBD Scenario Aim	To ensure the candidate is competent in taking informed consent from a patient.
CBD Focus (please provide the codes of the module(s) this scenario addresses)	*SLS424*　　　　*SLS425*

GSP Domains covered (enter X to indicate all that apply)	**GSP 1**	x	**GSP 2**	x	**GSP 3**	x	**GSP 4**	X	**GSP 5**	

CBD Scenario description	You see a lady in clinic with a clinical diagnosis of PKD – (Polycystic Kidney Disease). She requests genomic testing for this, as she would like to have preimplantation genetic diagnosis. You notice that the standard test for PKD is now Whole Genome Sequencing, instead of a 'panel test'. What are the key considerations as a genetic counsellor when taking consent from your patient for a whole genome sequencing test (WGS) as compared with ordering a panel test or single gene test? What would you want to discuss with your patient?
CBD Scenario model answer/ assessor guidance Detailed guidance that will be available for the assessors. Include guidance on what kinds of behaviours, actions, comments should secure a pass. What should the assessor expect to see? Assessors will be asked	Trainees are expected to talk through the process of how they would take consent for WGS, what they would specifically need to discuss with the patient, and what are some of the important considerations and issues that can arise when consenting patients and their relatives. Trainees should include the following in their answer: • Implications for the patient. • Implications for family members and consideration of sharing results with others.

to plan questions in advance including links to trainee's IACC submission.	• Potential outcome of the test – i.e. No pathogenic variant/pathogenic variant and variant of unknown significance. • Possibility of incidental findings. • Turnaround time of testing. • Potential opportunities to be involved in research in the future. • Awareness of potential psychosocial issues involved with WGS. • Importance of respecting patient autonomy and confidentiality. • Data protection issues that patients bring up. Good: expected response with good level of detail and good awareness of psychosocial issues. Satisfactory: answer covering the above topics with limited detail. Needs development: most of the above topics mentioned, but some key areas missed. Below expectations: many of the above topics not addressed.
Trainee instructions Please include any specific information to be provided to the trainee as part of the CBD scenario	The focus of this scenario is the process of taking consent for WGS and the issues that need to be discussed with the patient. You are not expected to go into details about PKD. We do not expect you to have experience of doing the Record of Discussion as this will vary across centres.

Criteria being assessed by this CBD scenario

Aspect	Please indicate if this criterion is being assessed
1. Understands the clinical context of the scenario, including priority setting and testing strategies	X
2. Understands scientific principles of scenario	X
3. Can discuss the relevant procedures involved in the scenario and associated health and safety issues	
4. Understands and applies the appropriate test validation, IQC, EQA, relevant professional/clinical guidelines	X
5. Understands and applies associated IT/bioinformatics and other appropriate resources	
6. Is able to interpret and report patient results and provide appropriate clinical advice	
7. Can discuss the significance of patient results within the clinical context of the referral	X
8. Understands the ethical, legal and social implications of the scenario	X

9. Is aware of the importance of audit and can use this tool effectively	
10. Output meets accepted laboratory/professional standards	
11. Demonstrates awareness of the limits of responsibility and when to seek advice	
12. Consideration of patient/professionalism	
13. Overall ability to perform	