National School of Healthcare Science

NHS England

IACC 2023 Case-based discussion (CBD) scenario

Specialty:	**Cancer Genomics**

CBD Scenario

CBD Scenario Title	Cancer Genomic testing for patients with melanoma										
CBD Scenario Aim	To demonstrate an understanding of standard of care (SoC) testing for patients with melanoma, including a brief overview of laboratory methodologies and a demonstration of how the results of genomic testing impact patient diagnosis and treatment.										
CBD Focus (please provide the codes of the module(s) this scenario addresses)	*SLS428*										
GSP Domains covered (enter X to indicate all that apply)	**GSP 1**		**GSP 2**	X	**GSP 3**	X	**GSP 4**		**GSP 5**		
CBD Scenario description	A 38-year-old Caucasian female with a history of extensive ‘tanning bed’ use has a longstanding mole on the right shin. A biopsy of the lesion and subsequent histology confirmed a diagnosis of a non-ulcerated superficial spreading melanoma [Breslow thickness 1.8mm (pT2a). **Histopathology on a subsequent sentinel lymph node biopsy confirms metastatic melanoma. Molecular genetic analysis was requested.** 1. Please describe the procedures that are followed to process a formalin fixed paraffin embedded (FFPE) tissue section from receipt in specimen reception to data analysis. 2. Please describe the testing strategy for a sample referred with a diagnosis of melanoma; what gene(s) would you analyse? 3. Analysis of an NGS DNA panel detected a BRAF variant; c.1799T>A p.(Val600Glu) [V600E]. No additional variants were detected in either the DNA or RNA panels. A) What are the clinical implications for the patient?										

	B) What information would you include in the final report?
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 to plan questions in advance including links to trainee's IACC submission.	Histopathology on a subsequent sentinel lymph node biopsy confirms metastatic melanoma. Molecular genetic analysis was requested. 1.Please describe the procedures that are followed to process a formalin fixed paraffin embedded (FFPE) tissue section from receipt in specimen reception to data analysis. • Booking in process • Extraction process – DNA (and RNA) from paraffin embedded tissues • Sequencing methodology – Sanger sequencing (if rapid result is required), large panels vs targeted panels • Bioinformatics pipeline and data storage • Variant interpretation – variant interpretation software and/or manual analysis 2.Please describe the testing strategy for a sample referred with a diagnosis of melanoma; what gene(s) would you analyse? • BRAF codon 600 targeted analysis may be described • NGS DNA panel testing; BRAF, NRAS, KIT • Structural variants (RNA panel or FISH): NTRK1,2,3 • Copy number analysis by FISH or microarray may be requested for equivocal cases MYB, RREB1, CCND1, MYC, CDKN2A 3.Analysis of an NGS DNA panel detected a *BRAF* variant; c.1799T>A p.(Val600Glu) [V600E]. No additional variants were detected in either the DNA or RNA panels. A) What are the clinical implications for the patient? B) What information would you include in the final report? • Recognize that this is a common BRAF variant in melanoma • Interpret that the patient is likely to respond to BRAF/ MEK - targeted therapy. • KIT: No actionable variants detected • NRAS: No actionable variants detected • NTRK1, 2, and 3: No actionable variants • Class 1 BRAF variant – describe guidelines used (AMP, etc.)
Trainee instructions Please include any specific information to be provided to the trainee as part of the CBD scenario	Please give this information before the first question:

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	X
4. Understands and applies the appropriate test validation, IQC, EQA, relevant professional/clinical guidelines	
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	X
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	
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	