Genomic Medicine in Practice.

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Objectives

- Understand the role of the CNS in molecular testing
- The importance of molecular testing.
- How to request molecular testing
- Challenges around molecular testing
- Case study

Word cloud

What is your role and involvement in genomics?

The role of the CNS in Genomics

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- Ensuring the patient is well informed and is in agreement for testing.
- Ensuring colleagues understand the rationale for genomics
- Ensuring biopsies and molecular testing is arranged in a timely manner.

The importance of Molecular testing

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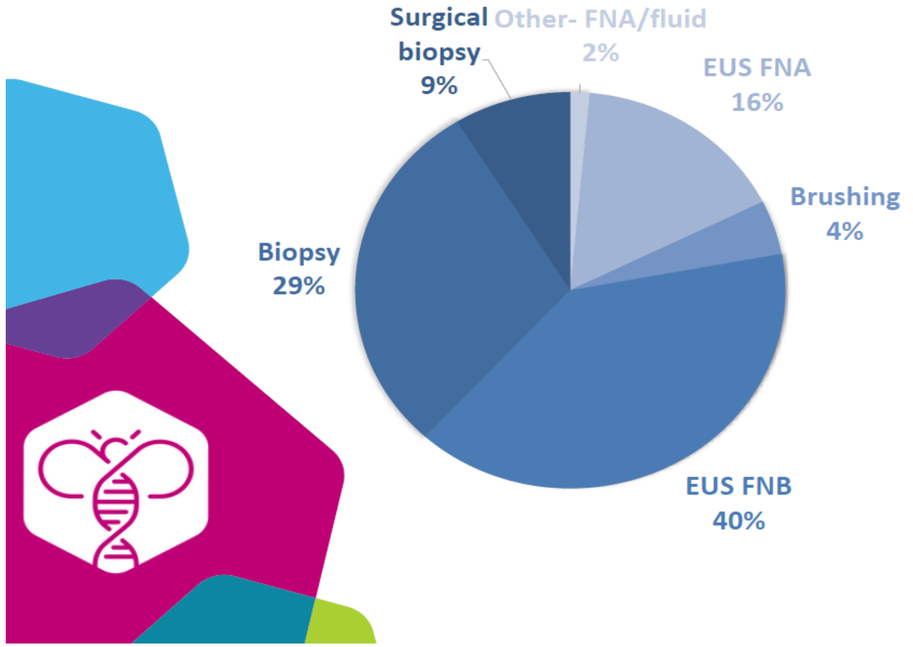
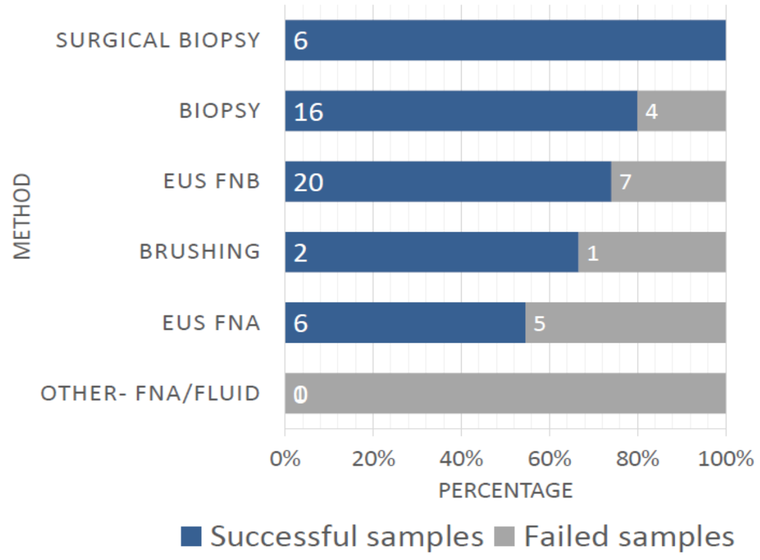
- Better treatment options for the patient
- Can avoid multiple invasive procedures
- Treatment can be considered earlier.

Tissue is the issue!! Pancreas





Denise Ng

How to request

- Molecular testing can only be requested once initial biopsy has been reported.
- Access your local genomic laboratory hub website.
- Complete testing request form.
- This can take up to a number of weeks for the report to be finalized.
- The report will be sent to the referrer.
- It is the responsibility of the referrer to forward the information to relevant parties.

Genitourinary/Renal/Pancreatic Tumour Test Request Form

UKAS MEDICAL 9865

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Patient Details		Payment Status: ☐ NHS ☐ Private	Referring Clinician	
Surname:			Consultant (in full):	
Forename:			Hospital (in full):	
DoB:	NHS No:		Department:	Tel:
Sex:	Hospital No:		Email:	
Address/Postcode:			Copy report to (if applicable):	

CLINICAL DETAILS

Pathology Laboratory Hospital/Trust:

PLEASE INCLUDE A COPY OF THE PATHOLOGY REPORT

Pathology block/sample no.:

Sampling Date:

PLEASE COMPLETE AND FORWARD TO THE PATHOLOGY LABORATORY HOLDING THE SAMPLE.

CI Code*	Clinical Indicaticn Name	Test Name	Test Code	Please tick
M18	Renal Cell Carcinoma - Adult	FH, SDHA, SDHB, SDHC, SDHD, VHL, ELOC (TCEB-1), TSC1/2, MET, BRAF	M18.2	
		TFE3, NTRK fusions	M18.6	
M217	Bladder Cancer	FGFR2, FGFR3	M217.1	
		FGFR2, FGFR3, NTRK fusions	M217.3	
M218	Prostate Cancer	BRCA1, BRCA2	M218.1	
		NTRK fusions	M218.2	
M219	Pancreatic Cancer	BRCA1, BRCA2	M219.1	
		NTRK fusions	M219.2	
		MSI Testing	M219.5	
Various	Any Tumour Type	NTRK fusions	Various	

*For full details of genes covered see national genomic cancer test directory (https://www.england.nhs.uk/publication/national-genomic-test-directories/).
NB For WGS,FISH,CNS and ctDNA testing please see: https://mft.nhs.uk/nwglh/test-information/cancer/solid-tumour/sample-requirements/referral-form/

PATHOLOGY LABORATORY – please complete

Please note 2 tubes of curls are required for all testing. *For sample requirements please see reverse or https://mft.nhs.uk/nwglh/*

Please circle or state the approximate neoplastic cells (%) in the sample sent for analysis *(this information is important in reducing the risk of false negative results).*

1-5#	6-10#	11-20#
20-50	50-75	>75

Overall neoplastic cell content _____% #*Neoplastic cells in marked area _____%*

#Where overall neoplastic cell content <20% and macrodissection would enhance % of neoplastic cells, please send slide mounted sections with corresponding marked H&E stained slide.

Efficacy of target medicine

SOC chemotherapy (FOLFOX)

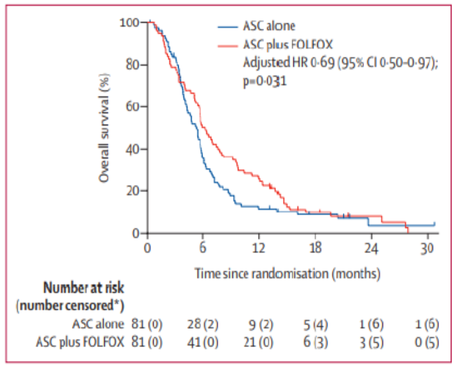
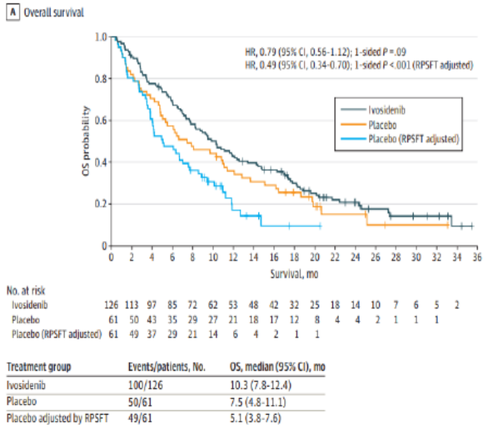


Figure 2: **Overall survival**
The HR is adjusted for the three stratification factors (platinum sensitivity, serum albumin concentration, and disease stage). ASC=active symptom control. FOLFOX=folinic acid, fluorouracil, and oxaliplatin. HR=hazard ratio. *Numbers are cumulative.

FOLFOX
Median overall survival 6.2 months

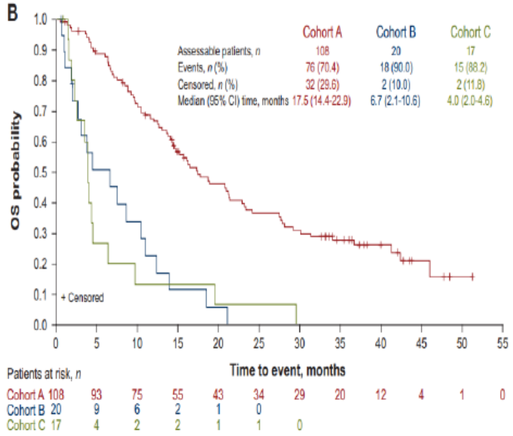
IDH1 mutation



Treatment group	Events/patients, No.	OS, median (95% CI), mo
Ivosidenib	100/126	10.3 (7.8-12.4)
Placebo	50/61	7.5 (4.8-11.1)
Placebo adjusted by RPSFT	49/61	5.1 (3.8-7.6)

Ivosidenib
Median overall survival 10.3 months

FGFR2 fusion



Pemigatinib
Median overall survival 17.5 months

1) Second-line FOLFOX chemotherapy versus active symptom control for advanced biliary tract cancer (ABC-06): a phase 3, open-label, randomised, controlled trial Lamarca, Angela et al. The Lancet Oncology, Volume 22, Issue 5, 690 – 701
2) Final Overall Survival Efficacy Results of Ivosidenib for Patients With Advanced Cholangiocarcinoma With IDH1 Mutation: The Phase 3 Randomized Clinical ClarIDHy Trial. Zhu AX, Macarulla T, Javle MM, et al. JAMA Oncol. 2021, 7(11):1669–1677
3) An open-label study of pemigatinib in cholangiocarcinoma: final results from FIGHT-202☆Vogel, A. et al. ESMO Open, Volume 9, Issue 6, 103488

Challenges

- Procedural priority
- Patient fitness
- Timing of MDT discussions
- Cytology V Biopsy
- Delays
- Understanding by colleagues

Case Study

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- 63 Year old
- Good performance status
- Nil past medical History
- Presented jaundice
- Investigations shown a possible head of pancreas cancer
- Discussed with HPB on team, who felt potentially resectable
- MDT – Borderline resectable

Plan:

- Arrange ERCP + EUS to treat biopsy and take core biopsy
- Molecular testing completed and now under oncology care

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THANK YOU FOR LISTENING!