Prostate cancer – mainstream genomic testing

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Consultant Genetic Counsellor

Mainstreaming genomic testing

- Genetic/genomic counselling performed by non-genetic healthcare professionals
- More access to testing for patients who may benefit
- Testing earlier in the patient journey may alter the options that may be available

National Test Directory – R430 criteria

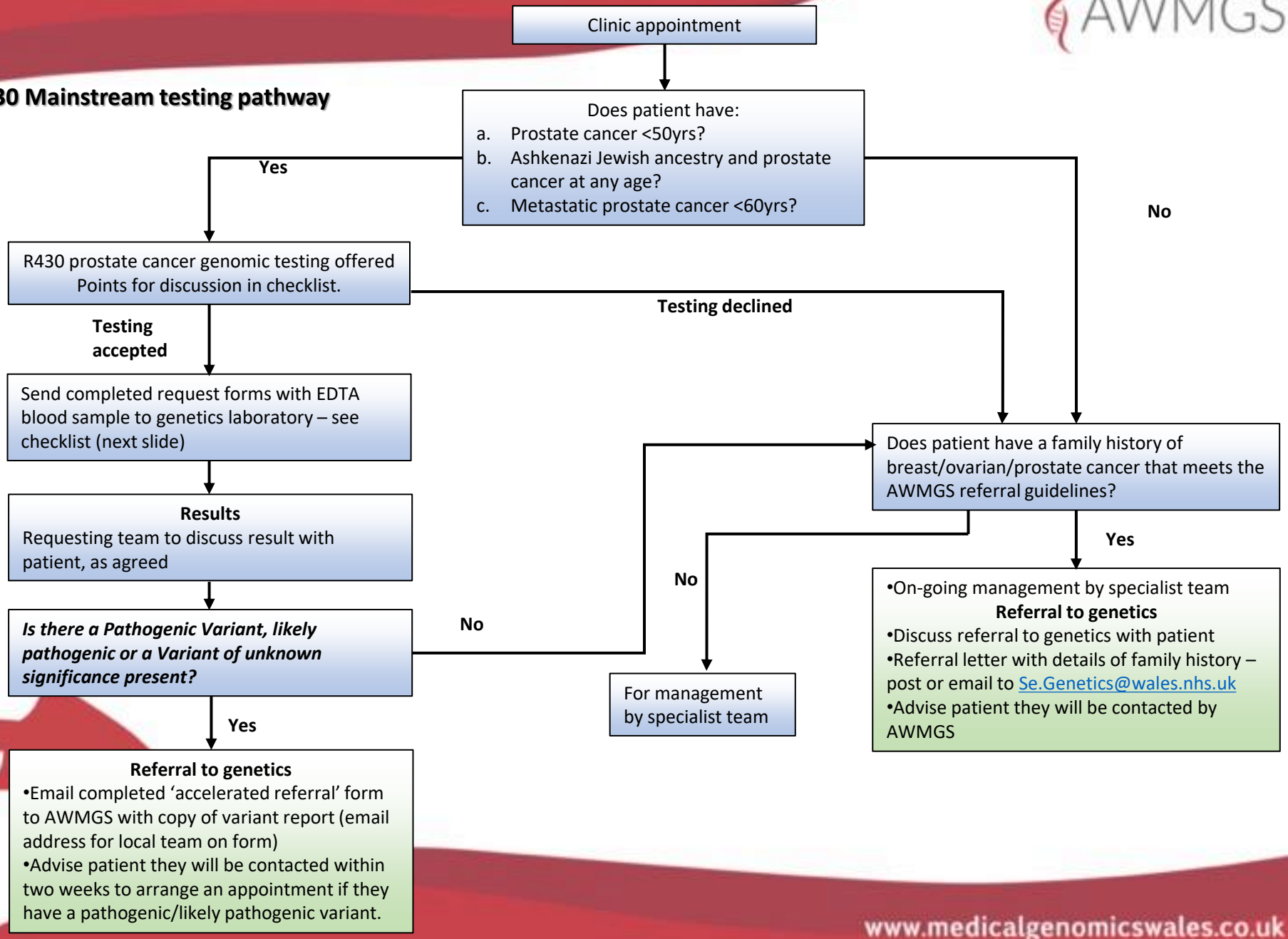
Mainstream

- Proband diagnosed with prostate cancer <50yrs
- Ashkenazi Jewish ancestry and prostate cancer at any age
- Proband diagnosed with metastatic prostate cancer <60yrs

Clinical Genetics

- Proband diagnosed with prostate cancer with a family history of prostate cancer, where estimated likelihood of identifying a pathogenic variant in the relevant target genes is at least 10%

R430 Mainstream testing pathway



M218.1/R444.2 – prostate cancer gene testing and PARP inhibitors

- If patient meets the criteria for PARPi, should use M218.1/R444.2 indication first.
  - R430 will only look for germline variants

R444.2 Prostate Cancer
Metastatic, castration-resistant prostate cancer where somatic tumour testing (M218.1) has failed.

Prostate cancer genes – current R430 panel

Gene	Associated cancers
BRCA1	Breast & ovarian
BRCA2	Breast, ovarian, prostate, pancreatic
PALB2	Breast, pancreatic, ovarian
CHEK2*	Breast, prostate, colorectal
ATM*	Breast, prostate, other risks undefined
MLH1	Lynch syndrome
MSH2	Lynch syndrome
MSH6	Lynch syndrome

Patient discussion

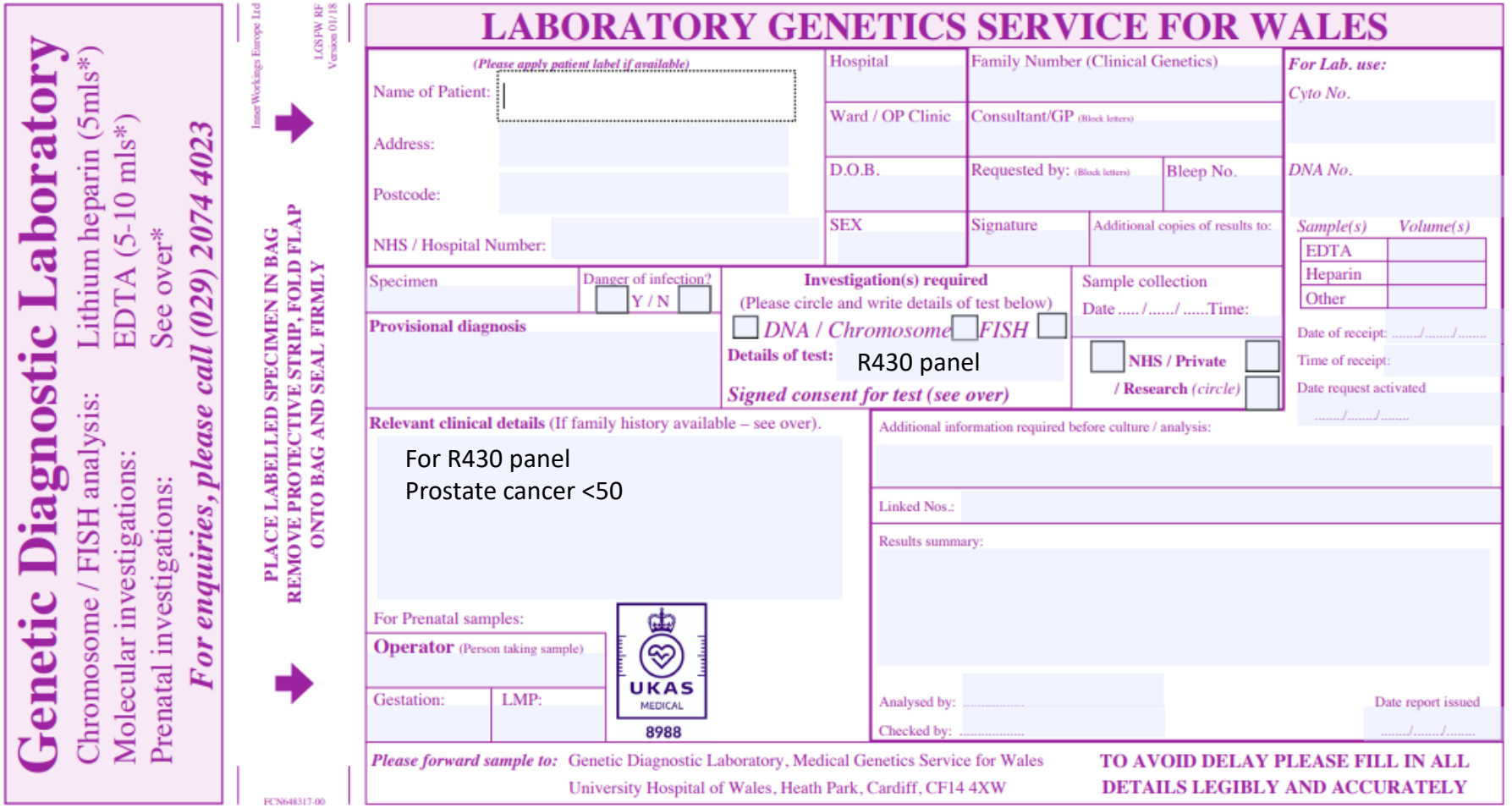
- Reason for offering test
  - Including potential implications for relatives
- What does the test involve?
  - Including turn around time
- Possible outcomes
- Potential implications for patient
- Turnaround time for results

What does the test involve?

- Blood sample – one 4ml EDTA tube is sufficient
  - Include on the test request form the R number for the panel – R430
  - Include the criteria the patient meets
- TAT for panel is currently 4 months

Currently the only formal agreement for testing to be expedited is where chemotherapy treatment must commence within a set timeframe

Lab request form



Genetic Diagnostic Laboratory

Chromosome / FISH analysis: Lithium heparin (5mls*)
Molecular investigations: EDTA (5-10 mls*)
Prenatal investigations: See over*

For enquiries, please call (029) 2074 4023

InnerWorkings Europe Ltd

LGSPW RF
Version 01/18

PLACE LABELLED SPECIMEN IN BAG
REMOVE PROTECTIVE STRIP, FOLD FLAP
ONTO BAG AND SEAL FIRMLY

FCN648317-00

LABORATORY GENETICS SERVICE FOR WALES

(Please apply patient label if available)

Name of Patient:

Address:

Postcode:

NHS / Hospital Number:

Hospital

Ward / OP Clinic

D.O.B.

SEX

Family Number (Clinical Genetics)

Consultant/GP (Block letters)

Requested by: (Block letters)

Bleep No.

Signature

Additional copies of results to:

For Lab. use:

Cyto No.

DNA No.

Sample(s)	*Volume(s)*
EDTA	
Heparin	
Other	

Date of receipt: ...../...../.....

Time of receipt:

Date request activated

...../...../.....

Specimen

Danger of infection? ☐ Y / N ☐

Provisional diagnosis

Investigation(s) required

(Please circle and write details of test below)

☐ *DNA / Chromosome* ☐ *FISH* ☐

Details of test: R430 panel

Signed consent for test (see over)

Sample collection

Date ...../...../.....Time:

☐ **NHS / Private** ☐

/ Research *(circle)* ☐

Relevant clinical details (If family history available – see over).

For R430 panel
Prostate cancer <50

Additional information required before culture / analysis:

Linked Nos.:

Results summary:

For Prenatal samples:

Operator (Person taking sample)

Gestation: LMP:

UKAS MEDICAL
8988

Analysed by: .....

Checked by: .....

Date report issued

...../...../.....

Please forward sample to: Genetic Diagnostic Laboratory, Medical Genetics Service for Wales
University Hospital of Wales, Heath Park, Cardiff, CF14 4XW

TO AVOID DELAY PLEASE FILL IN ALL DETAILS LEGIBLY AND ACCURATELY

Possible outcomes

- Pathogenic/likely pathogenic variant
  - Likely explanation for patient's diagnosis +/- family history
- No variants detected
  - Does not completely exclude there being a genetic explanation for patient's diagnosis +/- family history
  - If patient meets AWMGS referral criteria, referral should be offered
- Variant of unknown significance
  - Variant identified in a gene, but laboratory are unable to clarify whether it would explain patient personal +/- family history

Potential implications for relatives

- Testing for relatives
  - Only if a pathogenic/likely pathogenic variant detected
- Risk management options
  - Screening may be recommended depending on family history
  - Risk-reducing surgery may an option
- Referral to Clinical Genetics
  - If patient meets AWMGS referral criteria (as per pathway)

GeNotes - GeNotes: Genomic notes for clinicians | GEP | Health Education England | NHS (hee.nhs.uk)

- In Clinic
  - Short scenarios that guide clinicians through testing process

- Knowledge Hub
  - Concepts
  - Condition specific information

Genomics - CPD

- Introduction to Genomics in Healthcare – HEIW
  - Y Ty Dysgu e-learning platform – free to NHS staff
  - ytydysgu.heiw.wales
- Genomic Medicine modules

- Genomics of common and rare inherited diseases
- Application of genomics in infectious disease
- Omics techniques and technologies and their application to genomic medicine
- Molecular pathology of cancer and application in cancer diagnosis, treatment and monitoring
- Introduction to the counselling skills used in genomic medicine
- Bioinformatics, interpretation and data quality assurance in genomic analysis

www.medicalgenomicswales.co.uk