

CELL

NUCLEUS

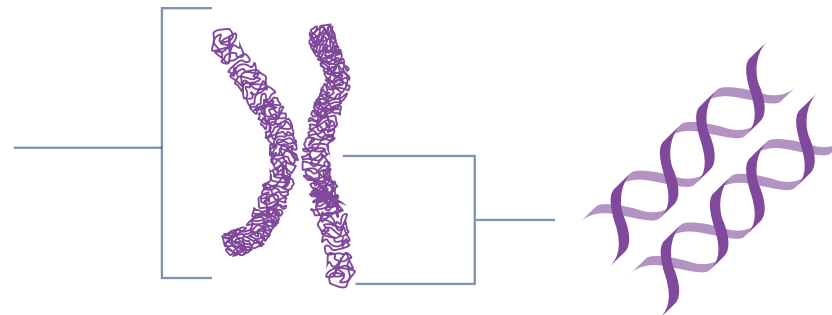
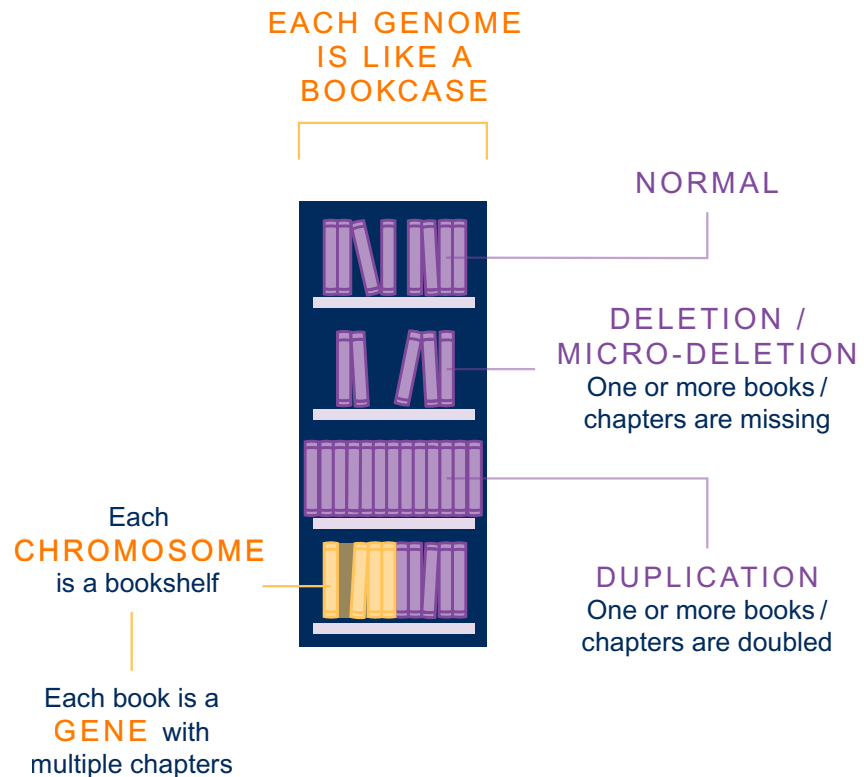
CHROMOSOME

GENE

DNA

MICRO-ARRAY *high-level testing*

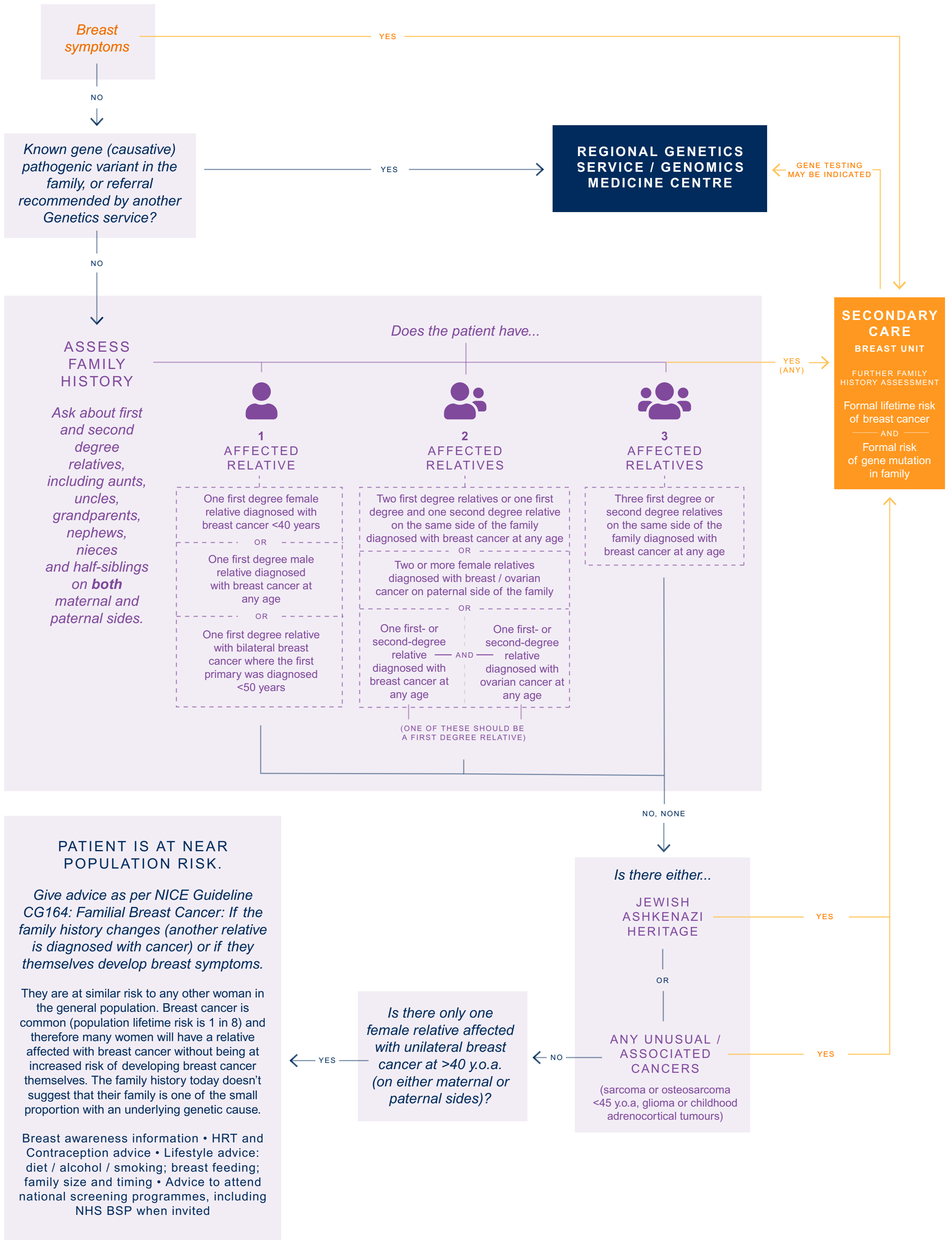
MICRO-ARRAY TESTING
IS TESTING AT THE
CHROMOSOME OR
BOOKSHELF LEVEL



CHROMOSOME

look at one section of
chromosome, not at
specific genes

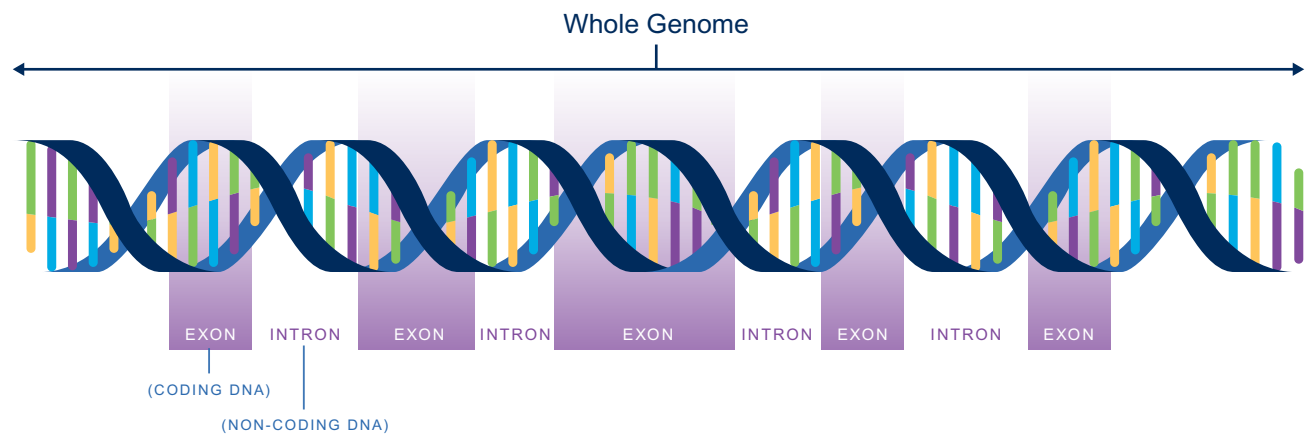
PATIENT PRESENTING WITHOUT A PERSONAL HISTORY OF BREAST CANCER



DIAGNOSTIC

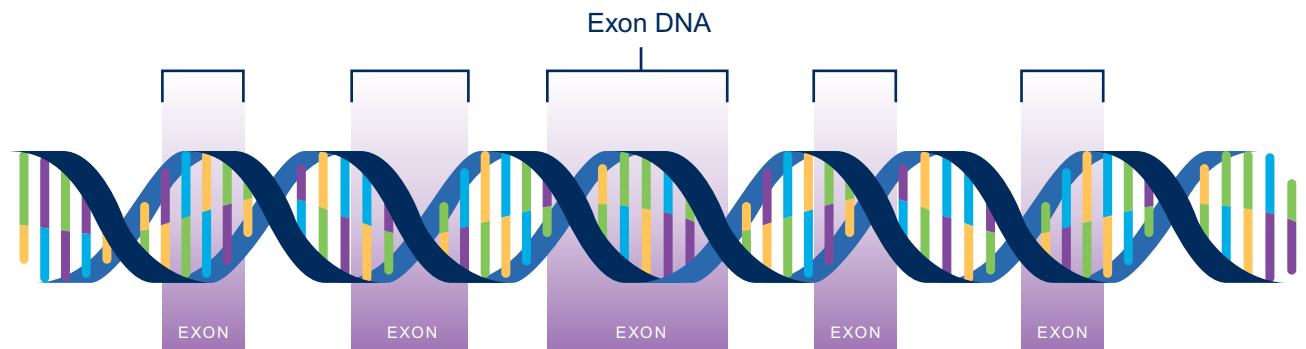
WHOLE GENOME

Testing whole genome, including coding & non-coding



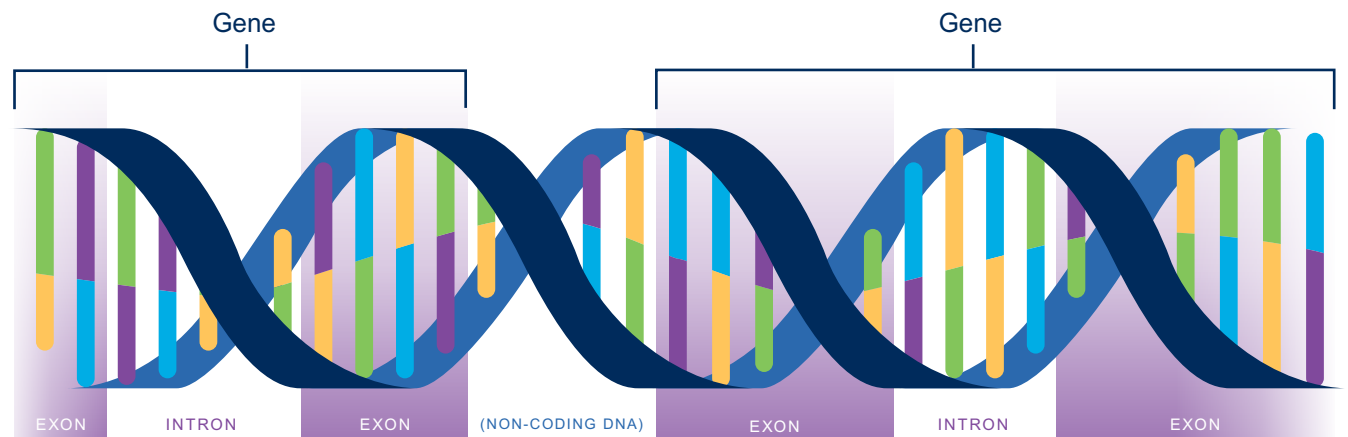
WHOLE EXOME

Testing whole genome, but only coding or exon DNA



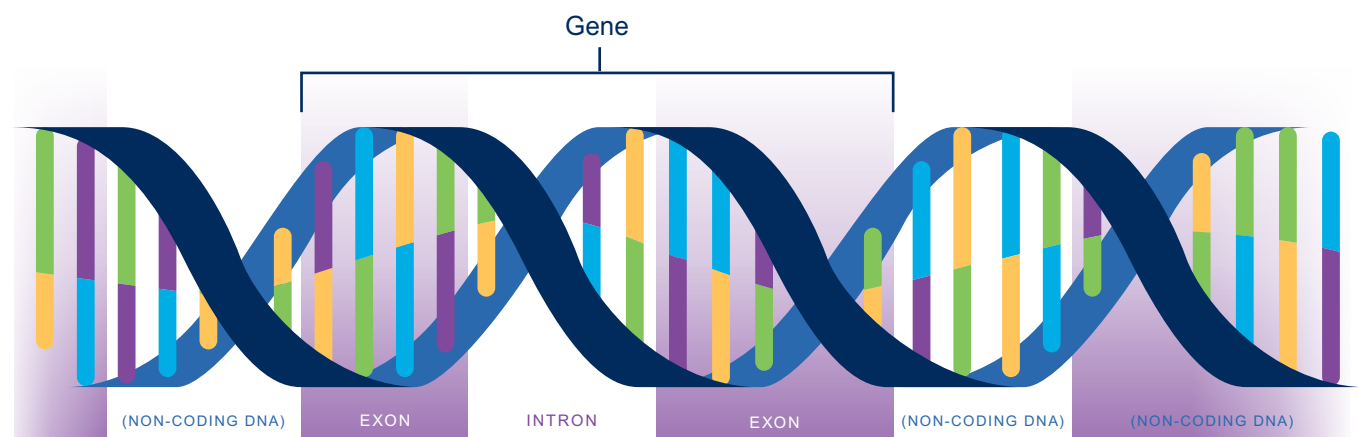
TARGETED PANEL

Testing one or multiple genes (up to 100) in coding & some non-coding DNA



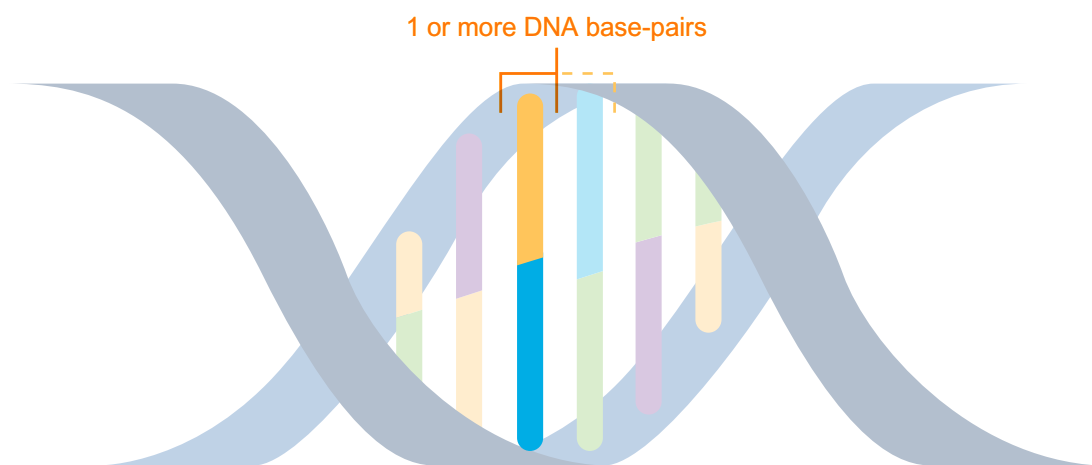
SINGLE GENE

Testing coding & some non-coding DNA for one single gene



PREDICTIVE

Targeted testing for genetic alteration (1 or more base pairs) within a gene which has already been identified in another family member e.g. a change in a single base pair resulting in variant BRCA1



SOMATIC DNA TESTING

Somatic DNA variants occur after conception, and can occur in any cell in the body except germ cells.

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Somatic DNA variants are therefore not inherited, and cannot be passed to offspring.

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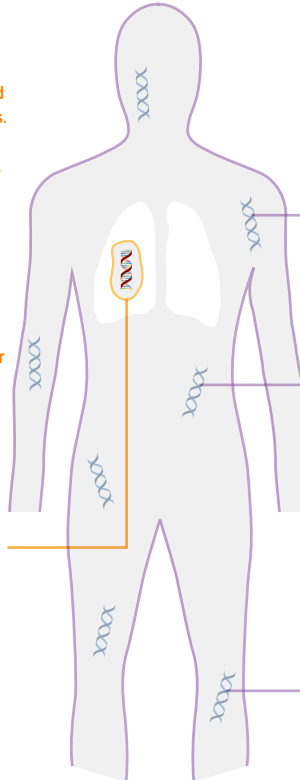
Somatic DNA variants may (but do not always) cause cancer: Somatic DNA testing is generally used to test tumour tissue, specifically to guide cancer treatment

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Examples would be HER2 testing in breast cancer when considering treatment with Herceptin, or EGF-R testing in non-small cell lung cancer



TUMOR



CONSTITUTIONAL DNA TESTING

Constitutional DNA is DNA found in every cell of the body, including germ cells, i.e. egg or sperm. Also called germ-line DNA as it is the source of DNA for all cells in the body.

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Constitutional DNA may be passed to offspring.

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Constitutional DNA testing (usually via blood sample) tests for inherited disorders.

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Examples would be testing for BRCA1 or BRCA2 gene variants which would confer a high risk of developing breast and ovarian cancer in women.

NHS GENOMIC MEDICINE SERVICE

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