Royal College of General Practitioners

Genomics Phrasebook

Genetics: single gene disorders

Genomics study of the entirety of DNA, the genome, together with the technologies which allow sequencing, interpretation and analysis

Genomic Medicine: application of genomics to clinical care

Gene: a segment of DNA that contains the biological instructions for a particular polypeptide, usually specific protein or component of a protein

- 'A recipe / instruction / computer programme for something about us e.g. eye colour, hair colour'

Pathogenic variant: an alteration in genetic sequence that increases an individual's susceptibility or predisposition to a certain disorder

- 'Change / spelling mistake / typo in a gene which explains your health problems'

Benign variant: an alteration in genetic sequence which is not disease-causing

- 'Change / spelling mistake / typo in a gene which doesn't cause any problems or harm'

Variant of unknown significance: an alteration in a genetic sequence whose association with disease risk is unknown

- 'Change / spelling mistake / typo in a gene which may explain your health problems or may be a chance finding, but currently, it's not possible to say whether it is significant or not'

Secondary finding (incidental finding): results which provide information about variants which are unrelated to the primary reason or clinical indication for testing

Additional looked-for finding: results which provide information about variants which are unrelated to the clinical indication for testing: patient opts in and consents to testing for these. These tend to be conditions with significant health implications, whose clinical course can be altered by screening and/or risk-reducing measures

Penetrance: the proportion of individuals with a particular genotype who express the associated phenotype / develop features of a condition

Diagnostic testing: genomic / genetic testing in someone affected with features of a condition to aid diagnosis

Predictive testing: genomic / genetic testing in an unaffected individual, specifically for a pathogenic variant known to be present in a family member