Genomics / Genetics in Healthcare

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What we are covering

- An introduction to genetics / genomics
- Information on the role of precision medicine in cancer and rare diseases
- Pedigree's (family history)
- GatewayC
- Examples of results from 100K Genome project
- Examples of genetic reports
- Pharmacogenetics
- Mechanism of Inheritance
- Online courses

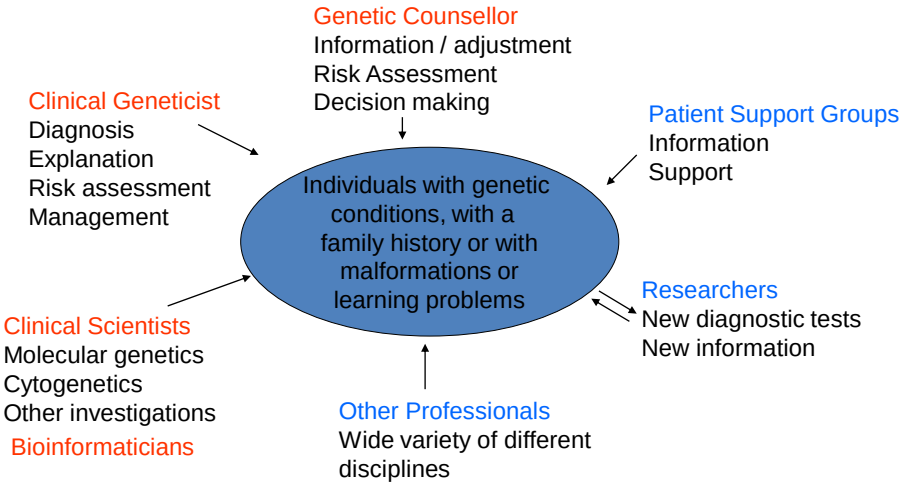
Genomics / Genetics

- Genetics is used in discussion of single gene diseases e.g. cystic fibrosis
- Genomics refers to a person's entire genetic code / DNA – their genome. Variations in different parts of the genome might increase or decrease the chances of developing many common multifactorial and polygenic diseases such as diabetes or heart disease.
- Genomic Medicine refers to the application of genomics to clinical care of patients; Regional Genetics Centres have now become Genomic Medicine Centres.

Genomic Medicine Centres

- Risk Assessment
- Surveillance
- Advice regarding risk-reducing measures including genetic counselling
- Consideration for genetic / genomic testing:
  - Single gene testing
  - Testing of multiple genes (gene panel)
  - Whole exome sequencing (WES)
  - Whole genome sequencing (WGS) ~100K Genome Project

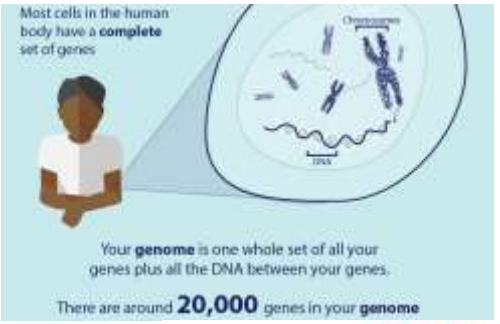
The Regional Genetic Team



Patient Questions

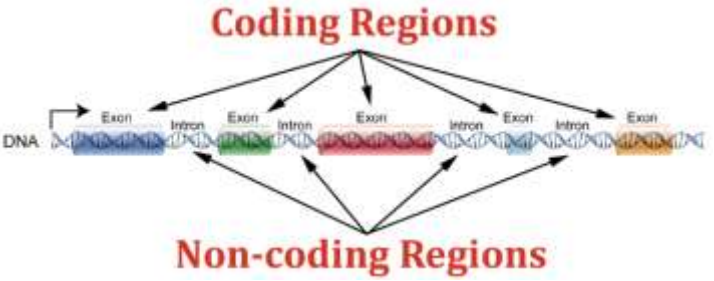
- Why did it happen?
- Can it be treated/cured?
- Am I to blame?
- What is the outlook for the future?
- Will it happen again?
- Are there implications for the rest of the family?

What Is A Genome?



Only 2% of DNA codes for proteins (the EXOME)

The other 98% contains some Important regulatory elements



Ways of looking at genes:

- The exome
- All ~20,000 genes
  - ~30-60,000,000 (30-60 million) bases

- The whole genome
  - All ~20,000 genes and the sequence in between.
  - ~2% is genes, 98% is not genes
  - 3,000,000,000 (3 billion) bases

MANCHESTER 1824
The University of Manchester

Genomics england

The GP's Role

- GPs are the frontline for personalised care
- Genomics provides another dimension for personalised care. This is being driven by:
  - Advances in genetic technology
  - Improved understanding of genomics amongst the medical and patient populations
  - Increasing demand genetic and genomic investigation from both doctors and patients
- We want GPs to be EMPOWERED to embrace, promote and effectively utilise genomic medicine

Rare disease diagnosis in Primary Care

- Individual disease frequency of <1/2000 of population
- This includes diseases we are familiar with such as Cystic Fibrosis
- Over 7000 rare diseases, affecting 1 in 17 of the UK population (>3 million people in the UK)
  - In a typical surgery of 8000 patients, ~470 patients have a rare disease
- Diseases often disabling, shorten life, costly
  - ~75% affect children
  - ~30% die by their 5th birthday
- ~ 85% are estimated to be caused by defects in a single gene

Collectively RARE DISEASES ARE NOT RARE

Rare disease diagnosis in Primary Care

- Although ~95% of rare diseases have no approved therapy, there are still benefits:
  - End uncertainty - “The Diagnostic Odyssey"
  - Explanation can help patients rationalise and obtain some closure
  - Access expert care, patient support groups.
  - Access educational and social support
  - Opportunities for clinical trials and drug development
  - Reproductive choice and family planning (genetic counselling)
  - Decisions about lifestyle – home / work etc.

The strategic approach -

tailoring treatment & management to a patient’s individual makeup

Now

- ‘One size fits all’ treatment based on symptoms
- Organ/ speciality organisation of services and professions
- Limited use of genomic and molecular markers
- Diagnostic and other clinical data not linked

2020

- New taxonomy of medicine based on underlying cause and personal response
- Comprehensive linked diagnostics to give a full picture of patient
- Tailored, more-effective therapies for better outcomes
- Integrated clinical services taking a ‘whole body‘ approach

Increasingly precision interventions based upon carefully identified subgroups within the broader population

Genomic diagnosis shaping precision management – neonatal diabetes

- mutations in >25 different genes cause neonatal diabetes
- new genomic technology has found 5 new genetic subtypes which inform therapy options





***KCNJ11* p.V59M** **Permanent diabetes and developmental delay**	***EIF2AK3* p.E371*** **Wolcott Rallison Syndrome**	***FOXP3* c.227delT** **IPEX syndrome**	***GATA6* c.1448-1455del** **Syndromic pancreatic agenesis**	***STAT3 p.T716M*** **Multi-organ autoimmune disease**
Sulphonylurea therapy	*Liver Transplant*	*Bone Marrow Transplant*	*Insulin and exocrine supplements*	*? STAT3 inhibitor*

Five babies; five different treatments

Rare Disease Summary

- Rare diseases are collectively common
- Genetics and genomics can end the diagnostic odyssey and inform management
- Genetics and genomics are becoming accessible, commonplace and integrated into clinical care
- Please do refer!

The role of the GP in cancer genomics

- Cancer is where GPs currently experience the greatest impact from genomics.
- GP's may have a potential role in identifying cancer that can be facilitated by family-history tools.
- Patients may be managed in primary care if they are at **near-population risk:**
  - **Example:** One 1st or 2nd degree relative presenting with unilateral breast cancer >40 years with no additional factors such as bilateral breast cancer, male breast cancer, ovarian cancer, paternal history breast cancer (2 or more relatives)
- Please do refer!

Possible scenarios in GP land.

- *My mother had breast cancer when she was only 47, and I am now 40. Can I have a mammogram? Her brother had it in his 60s but I don't think that's relevant as he is a man.*
- *My sister has had a BRCA1 gene test and carries a gene change. What does this mean for me and can I have a test too?*

Inherited Cancer Syndromes

Cancer syndrome/ lifetime risk of cancer for women	BRCA1	BRCA2	HNPCC)Lynch)
Breast	80%	60%	
Ovarian	40%	20%	10%
Endometrial			60%
Male breast cancer		Y	
Prostate		Y	
Pancreatic		Y	
Colorectal cancer			60-80%
Gastric			Y
GU tract			Y

Familial Cancer: red flags?

- Red flags for inherited cancer syndromes include:
  - Early onset
  - Bilateral
  - Associated with other malignancies
  - Known gene mutations (BRCA for example)
- Risk assessment: **Taking a family history in primary care is key**
- Risk stratification either as near-population or above-population risk within primary care is crucial to appropriate management

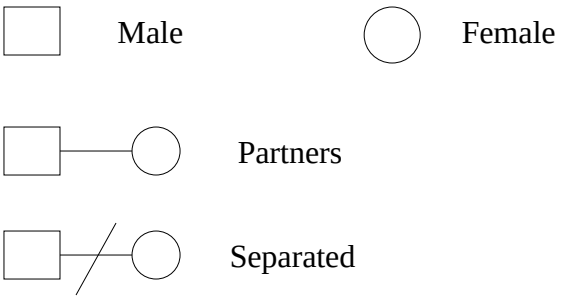
Drawing up a family pedigree

Hands on approx. 15 mins

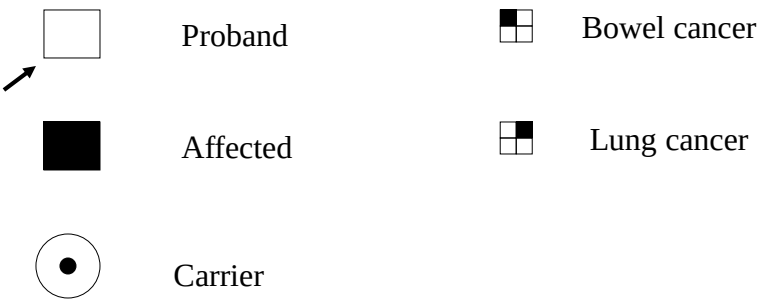
Why do we draw pedigrees

- Records family history
- May establish pattern of inheritance
- Aids diagnosis, investigation, management
- Use standard symbols

Symbols

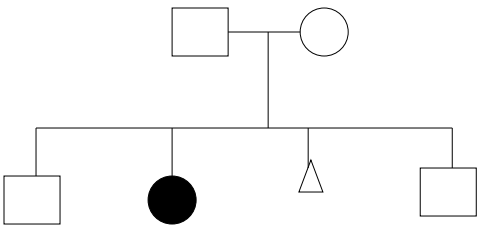


Symbols



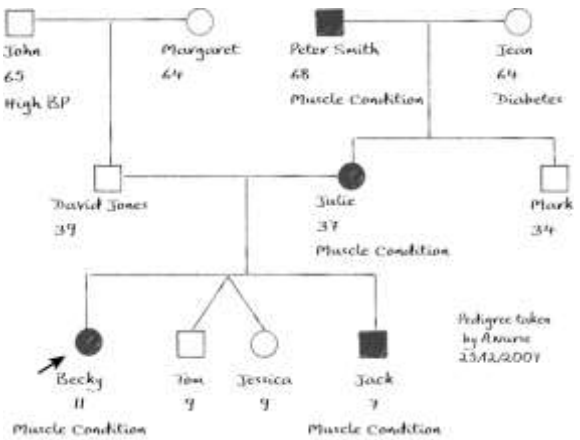
Male on left

Female on right



Oldest ⟶ Youngest

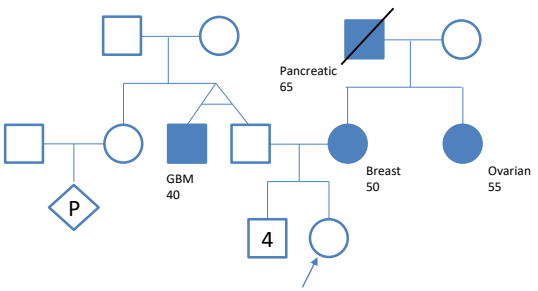
For example……



Exercise

Drawing a pedigree

Pedigree



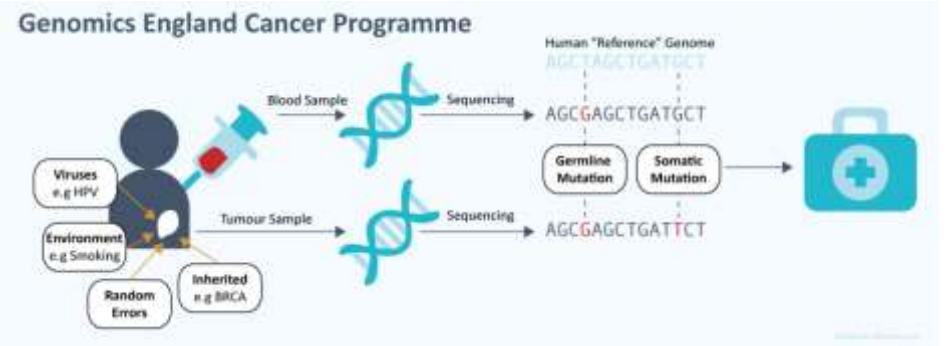
Germline Mutations

- These are mutations present in the sperm or the egg (the germline). These are passed on to the offspring at the moment of conception.
- These mutations are identified as they do not match to the human reference genome that we use for a guide.
- They are often passed down through generations, but can spontaneously occur in one generation (*de novo* mutation).
- They can affect your chances of developing cancer, for example changes in the BRCA1 and BRCA2 genes can lead to an increased risk of breast and ovarian cancers. We look for these changes if they are thought to be the cause of someone's cancer (if someone has red flags for example).

Somatic Mutations

- These are mutations that occur in the soma – cells that are not the sperm and the egg and instead stay only within the patient.
- These are occurring all the time (estimated 1-2 somatic mutations per cell division).
  - Some are intrinsic (errors in DNA replication for example)
  - Others are environmental and lifestyle (smoking and UV light are mutagens)
- Acquiring enough mutations in the key genes involve in cell replication or cell survival etc can lead to uncontrolled cell proliferation, manifesting as cancer.
- Somatic mutations can be identified by comparing genomes from the affected tissue (suspected cancer from a biopsy) with another, separate reference tissue (such as blood).
  - Can be sequenced through next-generation sequencing (WES, WGS)

Cancer Programme



GatewayC

- What is GatewayC?
- GatewayC is a FREE online cancer education platform for primary care professionals

across NHS England. It is designed to improve cancer outcomes by supporting earlier

diagnosis and the patient experience through:

  - Improved knowledge of symptoms
  - Increased confidence in when and when not to refer a patient
  - Improved quality of suspected referrals, reducing delays in the system
  - Improved communication to enhance the patient experience and support patients at each stage of their cancer pathway

Register today at gatewayc.org.uk/register

GatewayC

- How to register for GatewayC's FREE online cancer courses
  - Visit gatewayc.org.uk/register
  - Fill out the form and click submit
  - The GatewayC team will process your registration and email you with your new login details
  - Visit GatewayC to log in with your new details and start your online learning

Summary

- Around 5% of cancer results from an inherited cancer syndrome
- Risk assessment: family history / refer to Genomics Medicine centre if:
  - Clustering of any cancer in a family is a red flag e.g. 3 relatives with same cancer >60 yoa, 2 relatives <60 yoa
  - Known gene mutation in family or advised by another genetics service
  - 2 or more cancers: ovarian and / or associated cancers
  - Associated cancers: colorectal, endometrial, other GI (gastric, small bowel), GU tract
- Common management principles: surveillance, risk-reduction / lifestyle advice, symptom awareness, review if family history changes
- Genomic testing is advancing knowledge of familial cancer and inherited cancer syndromes and advancing targeted treatment for cancer
- Increasing access to genomic testing will impact on primary care

Useful information

- For information on genomics in primary care:
http://www.rcgp.org.uk/clinical-and-research/our-programmes/innovation/genomics-in-medicine.aspx

- For information on genomics education:
https://www.genomicseducation.hee.nhs.uk/

Information for patients:
- www.macmillan.org.uk
- www.cancerresearch.org
- www.ovarian.org.uk
- www.breastcancergenetics.co.uk

- NICE CKS

Useful links

- Clinical guideline [CG164] (breast cancer testing in families)

- https://www.genomicsengland.co.uk/about-genomics-england/the-100000-genomes-project/
- https://ukgtn.nhs.uk/
- https://www.nhs.uk/conditions/predictive-genetic-tests-cancer/
- https://www.cancerresearchuk.org/about-cancer/causes-of-cancer/inherited-cancer-genes-and-increased-cancer-risk/inherited-genes-and-cancer-types#inherited_genes0

- https://www.mangen.co.uk/healthcare-professionals/manchester-genetic-diagnostic-laboratory/cancer-genetics/

- RGCP Genomics Toolkit
- https://www.rcgp.org.uk/clinical-and-research/resources/toolkits/genomics-toolkit.aspx?utm_campaign=Programme+News+-+November+2019&utm_source=emailCampaign&utm_medium=email&utm_content=

- RGCP Position Statement on Direct to Consumer Genomic Testing
- https://www.rcgp.org.uk/policy/rcgp-policy-areas/genomic-position-statement.aspx

Results 100K Genome Project

- During the initial pilot study we recruited 600 patients to the 100K Genome Project.
- Started to get Whole Genome Sequencing results back,
- Example of positive finding from the results:
- A ten year old girl was hospitalised (on HDU) with a severe reaction to chickenpox, with a history of severe chest infections.
- Previously tested for known immunodeficiency genes, but nothing was found.
- Results from this study identified an extremely rare mutation previously reported to be related to immunodeficiency in children from 5 families.
- Three of these died as a result of overwhelming infections.
- Based on this result the child will be given a stem cell transplant. The child's sister was also tested and found to also carry this mutation, and will also receive a stem cell transplant.
- This mutation has been added to the gene panel for testing for immunodeficiency in children, changing the face of clinical practice.

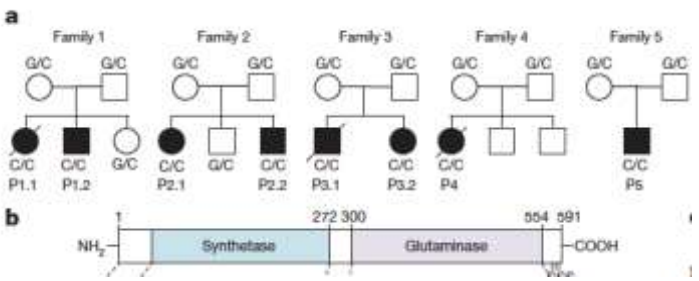
Homozygous variant in *CTPS1* c.1692-1G>C
Known splicing mutation

LETTER

doi:10.1038/nature13186

CTP synthase 1 deficiency in humans reveals its central role in lymphocyte proliferation

Emmanuel Martin[1,2], Noé Palmic[1,2], Sylvia Sanquer[3], Christelle Lenoir[1,2], Fabian Hauck[1,2], Cédric Mongellaz[4], Sylvie Fabrega[2,5], Patrick Nitschké[2,6], Mauro Degli Esposti[7,8], Jeremy Schwartzentruber[9], Naomi Taylor[4], Jacek Majewski[9], Nada Jabado[9,10], Robert F. Wynn[7], Capucine Picard[2,11,12], Alain Fischer[1,2,13,14], Peter D. Arkwright[7]* & Sylvain Latour[1,2,3]*



Whole genome sequencing enables definitive diagnosis of Cystic Fibrosis and Primary Ciliary Dyskinesia

Understanding the genomic basis of inherited respiratory disorders can assist in the clinical management of individuals with these rare disorders.

We applied whole genome sequencing for the discovery of disease-causing variants in the non-coding regions of known disease genes for two individuals with inherited respiratory disorders.

We were able to pinpoint candidate non-coding variants within the non-coding genome and demonstrate aberrant RNA splicing as a result of deep intronic variants in *DNAH11* and *CFTR*. These findings confirm clinical diagnoses of primary ciliary dyskinesia and cystic fibrosis.

Clinical findings

Proband 1 - a late diagnosis of cystic fibrosis at 17 years old.
Genetic testing uncovered the common del508 mutation in *CFTR*. A sweat test was positive: sweat chloride 68 mmol/L (normal range = 0-39 mmol/L); sweat conductivity 92 mmol/L (normal range = 0-49 mmol/L). The proband is currently 51 years old but her disease severity has progressed and she is awaiting double lung transplant.

Proband 2 - diagnosed in childhood with bronchiectasis, she is currently 54 years of age. Nasal nitric oxide levels were extremely low at 4 parts per billion (*ppb*, normal range = <25 ppb) consistent with a diagnosis of PCD. Three examinations of the proband's cilia with electron microscopy (EM) showed a significant proportion of static and dyskinetic cilia with a high ciliary beat frequency: 20.2Hz (95%CI=19.8-20.5Hz); 22.0Hz (95%CI=20.1-23.2Hz); and 20.1Hz (95%CI=19.8–20.5Hz). EM histology showed normal dynein arms and microtubules with no ciliary disorientation, and conical ciliated protrusions were observed from epithelial cells. These findings are consistent with mutations in *DNAH11*. Genetic testing identified a heterozygous nonsense mutation in *DNAH11*.

Table 1. Step-wise genomic assessments for individuals with inherited respiratory disorders

Testing Strategy:	Appropriate technologies:	Testing for Proband 1:	Testing for Proband 2:
1. Assess established disease-causing mutations	Genotyping arrays Direct mutation screening	Direct mutation assessment, 14 mutations (Dec 1996) ***CFTR p.Phe508del het***	Elucigene, CF-EU2 kit (Mar 2011) ***negative***
2. Discover disease-causing mutations in protein coding regions of known disease genes	Gene panel DNA sequencing WES (with virtual gene panels)	MRC-Holland MLPA P091 (Mar 2013) ***negative*** CFTR exon sequencing (Mar 2013) ***CFTR p.Phe508del het***	Invitae, 31 gene panel (Mar 2016) ***DNAH11 p.Tyr2870Ter het***
3. Discover disease-causing mutations in non-coding and regulatory regions of known disease genes	WGS (with virtual gene panels)	UK 100,000 genomes project (Aug 2018) ***CFTR p.Phe508del het*** ***CFTR c. 3874-4522 A>G het***	UK 100,000 genomes project (Aug 2018) ***DNAH11 p.Tyr2870Ter het*** ***DNAH11 c.6547-963 G>A het***
4. Novel disease gene discovery	WES or WGS	n/a	n/a

WES, whole exome sequencing; *WGS*, whole genome sequencing; MLPA, multiplex ligation probe amplification; *het*, heterozygous

Genomics in Everyday Healthcare

- A Trial to Assess the Utility of a Pharmacogenetic Test to Avoid Aminoglycoside Induced Ototoxicity in Neonates

- Developing a Point-of-Care Pharmacogenetic Test to Avoid Antibiotic Related Hearing Loss in Neonates

- *Mitochondrial MT-RNR1* - m.1555A>G

The Problem

- Variant of known clinical interest: We have known since 1993 that individuals with m.1555 A>G develop profound irreversible sensorineural hearing loss if exposed to an aminoglycoside.
- Previous population based studies have estimated the prevalence to be approximately 1:500 (0.2%)
- The **aminoglycosides** are broad-spectrum, bactericidal **antibiotics** that are commonly prescribed for children, primarily for infections caused by Gram-negative pathogens. The **aminoglycosides** include gentamicin, amikacin, tobramycin, neomycin, and streptomycin.
- 90,000 babies per year in the UK are treated with gentamicin in Neonatal Units (NICE CG149): therefore **180 babies per year** could avoid deafness by using another equally efficacious antibiotic.

Option 1	**Option 2**
Use an Alternative Antibiotic for Every Child	**Routinely Test for the Variant**
Significant Concerns RE Antibiotic Resistance	**Current testing methods take ~2-5 days**

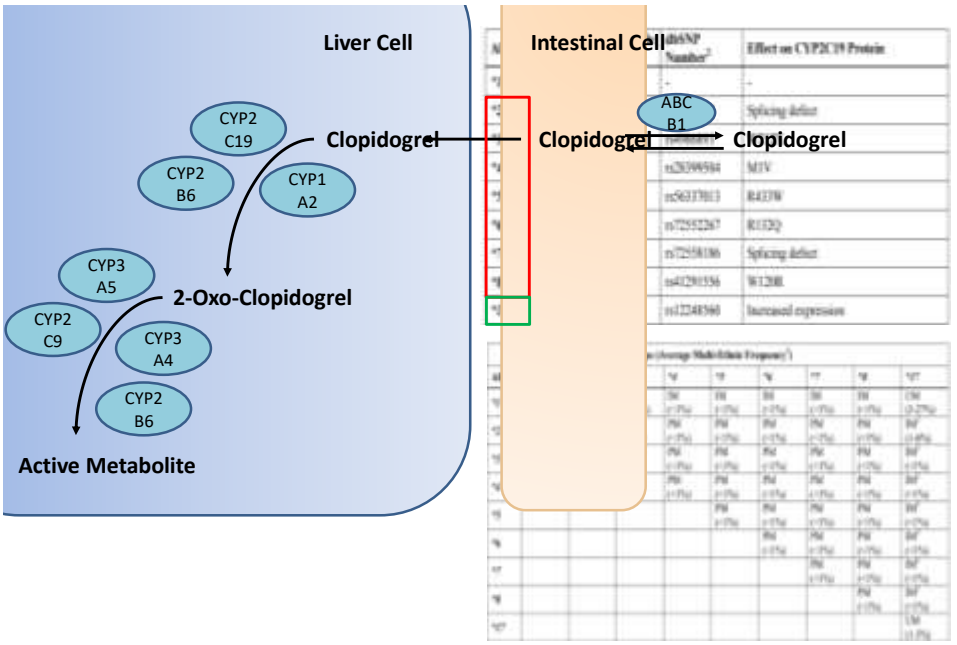
Methodology

genedrive

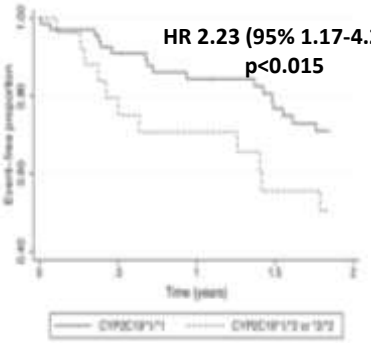
Outcome

- **Rapid:** Able to confirm the variant within a clinically relevant timeframe
- **Reliable:** Delivering consistent results comparable to gold standard sequencing
- **Intuitive:** Can be used by healthcare staff with minimal training 24/7
- **Portable:** Pont-of-Care, taking place by the bedside. No cold-chain
- **Non-Invasive:** Able to isolate the variant from buccal tissue
- **Inexpensive:** £60-£80 per assay. Cost of bilateral cochlear implants £61,000

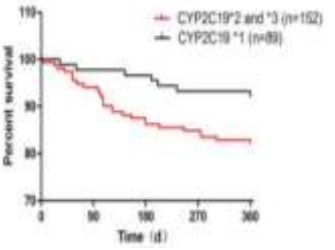
Optimising Antiplatelet Therapy in Stroke & TIA patients using Pharmacogenomics



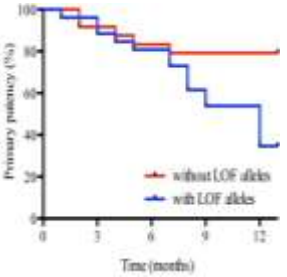
Recurrent Ischemic events in the Stroke Population



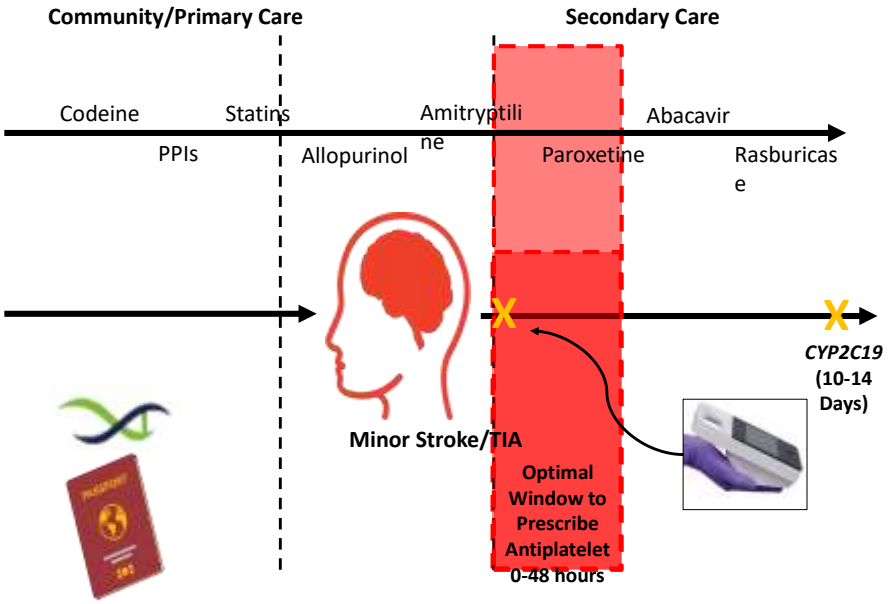
Mortality in Carotid Artery Stenting

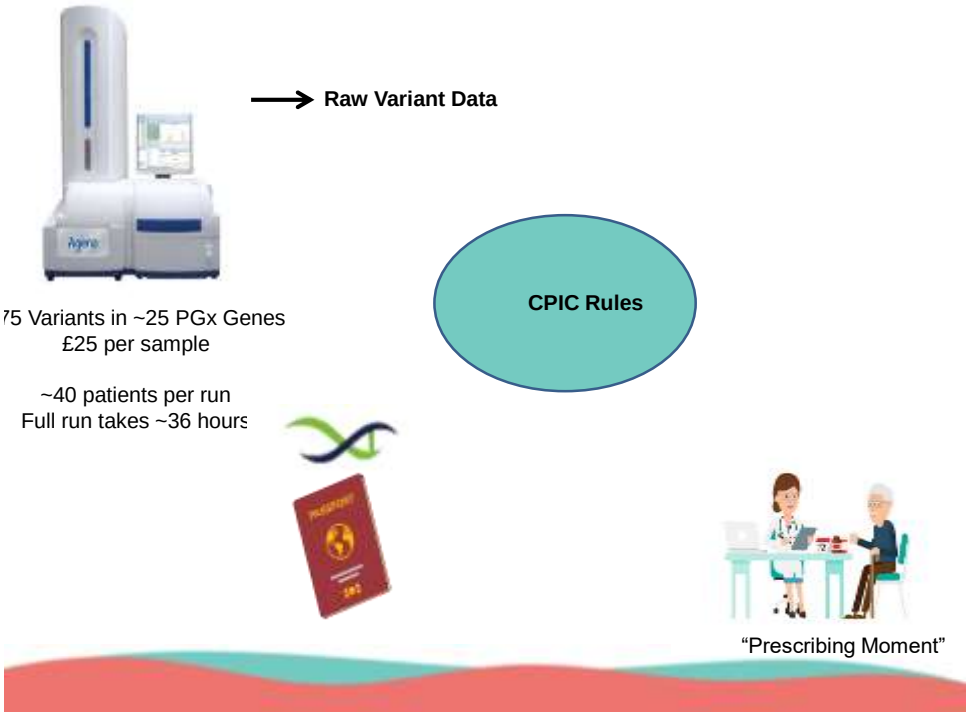


Patency of Endovascular Stents



1. Tornio et al. 2018. Investigating Real-World Clopidogrel Pharmacogenetics in Stroke Using a Bioresource Linked to Electronic Medical Records.
2. Wen et al. 2015. Association of *CYP2C19* Polymorphisms with the Clinical Efficacy of Clopidogrel Therapy in Patients Undergoing Carotid Artery Stenting in Asia.
3. Giugliano et al. 2016. Patients carrying *CYP2C19* loss of function alleles have a reduced response to clopidogrel therapy and a greater risk of in-stent restenosis after endovascular treatment of lower extremity peripheral arterial disease. Journal of Vascular Surgery


Community/Primary Care
Secondary Care
Codeine
Statins
Amitryptiline
Abacavir
PPIs
Allopurinol
Paroxetine
Rasburicase
X
X
CYP2C19
(10-14
Days)
Minor Stroke/TIA
Optimal
Window to
Prescribe
Antiplatelet
0-48 hours


Raw Variant Data
75 Variants in ~25 PGx Genes
£25 per sample
~40 patients per run
Full run takes ~36 hours
CPIC Rules
"Prescribing Moment"

Clinical Pharmacogenetics Implementation Consortium Guidelines for *CYP2C19* Genotype and Clopidogrel Therapy: 2013 Update

SA Scott[1], K Sangkuhl[2], CM Stein[3], J-S Hulot[4,5], JL Mega[6], DM Roden[7], TE Klein[2], MS Sabatine[6], JA Johnson[8,9,10] and AR Shuldiner[11,12]

Cytochrome P450 (CYP)2C19 catalyzes the bioactivation of the antiplatelet prodrug clopidogrel, and CYP2C19 loss-of-function alleles impair formation of active metabolites, further evidence from an expanded literature review. As in the 2011 guideline, recommendations for the use of other laboratory tests, such as platelet function monitoring, and cost-effectiveness

Phenotype to Genotype Table

Prescribing Guidelines

75 Variants in ~25 PGx Genes
£25 per sample

~40 patients per run
Full run takes ~36 hours

*CYP2C19 *1/*2*

Raw Variant Data → **Metaboliser Phenotypes**

Intermediate Metaboliser

CPIC Rules

Prescribing Advice

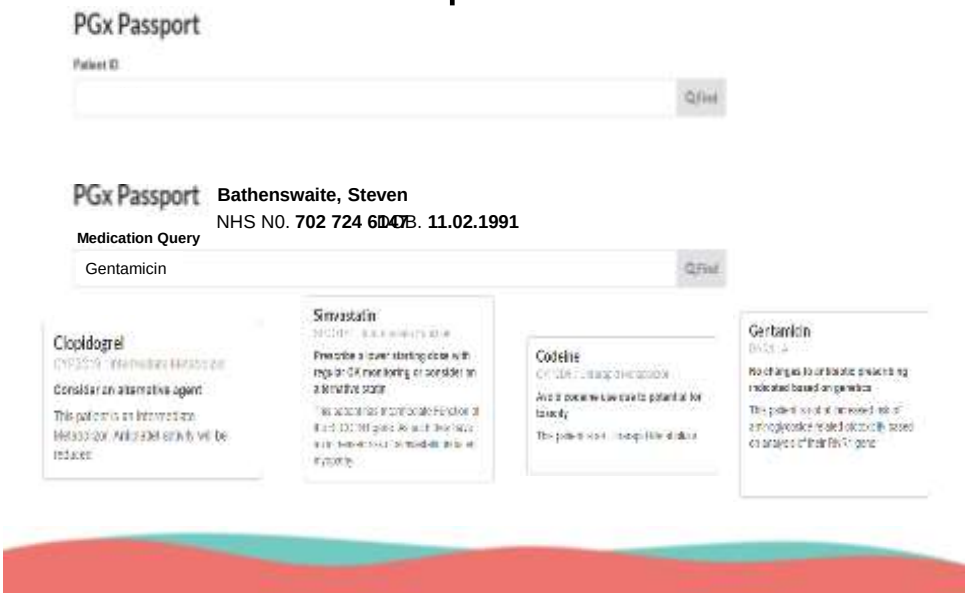
If IM consider alternative antiplatelet

Search PGx Passport by Drug Name

Clopidogrel → CYP2C19

"Prescribing Moment"

Greater Manchester PGx Passport



UKPGx Network Annual Open Meeting 2020 #UKPGx2020

7th Annual Open Meeting will take place at the Royal College of Physicians, London on March 10, 2020

http://www.uk-pgx-stratmed.co.uk/

#UKPGx2020

UK Pharmacogenetics & Stratified Medicine Network Annual Open Meeting

Exhibitors

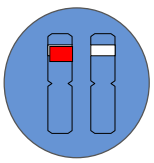
BRITISH PHARMACOLOGICAL SOCIETY

NHS Health Education England

illumina

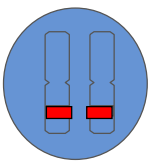
Yourgene Health

Mechanisms of inheritance



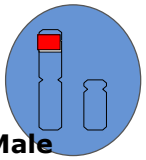
Dominant

Heterozygotes with **one copy** of the altered gene are affected



Recessive

Homozygotes with **two copies** of the altered gene are affected

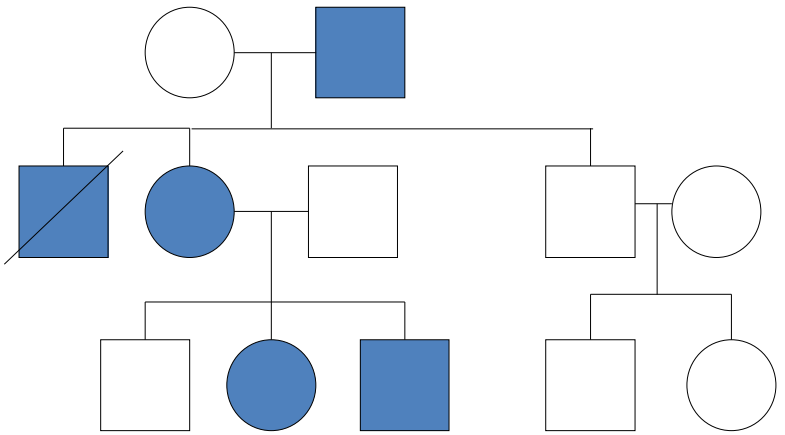


Male

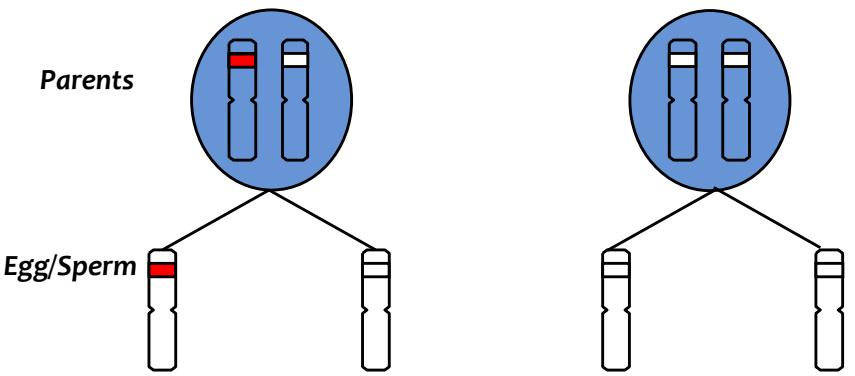
X-linked recessive

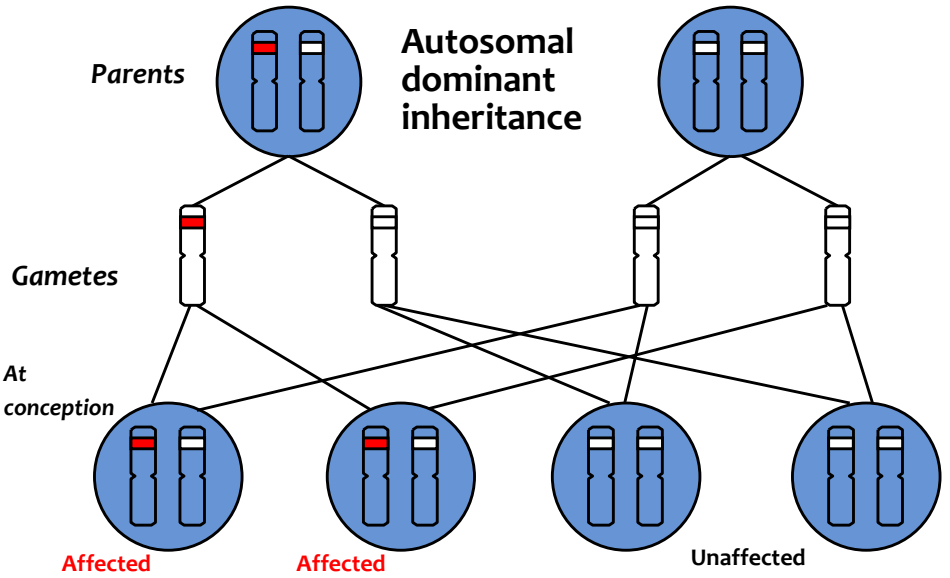
Males with **one copy** of the altered gene on the X-chromosome are affected

Autosomal Dominant



Autosomal dominant inheritance



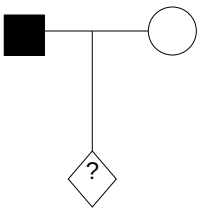


Autosomal Dominant disorders

- Heterozygotes largely or completely express the disorder
- Affected person often has an affected parent
- Affects either sex
- Transmitted by either sex
- 50% chance passing on the gene
- Variable expression
  - Manifestations or degree of severity vary from individual to individual and between families

Penetrance

- Penetrance = proportion of heterozygotes who show evidence of the effects of mutation.
- Expressed as %



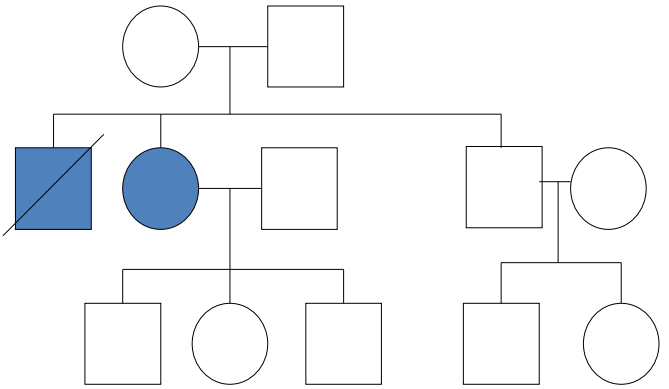
- May be age dependent eg HD, ADPKD

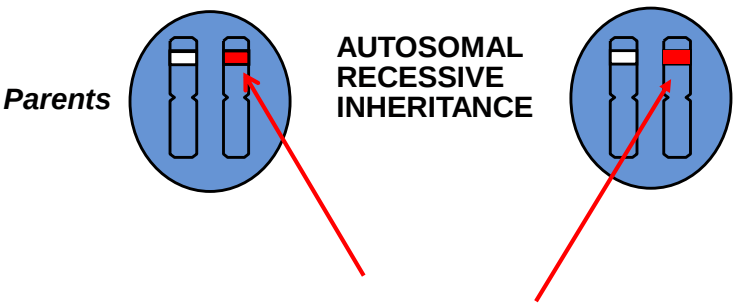
Risk = ½ x P

Autosomal dominant

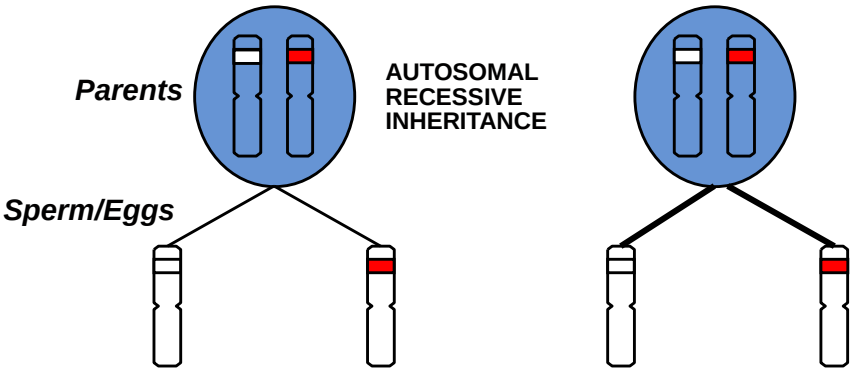
- Neurofibromatosis
- Huntington disease
- Achondroplasia
- Marfan syndrome
- Familial adenomatous polyposis coli
- Tuberous sclerosis
- Myotonic dystrophy
- Noonan syndrome

Autosomal Recessive





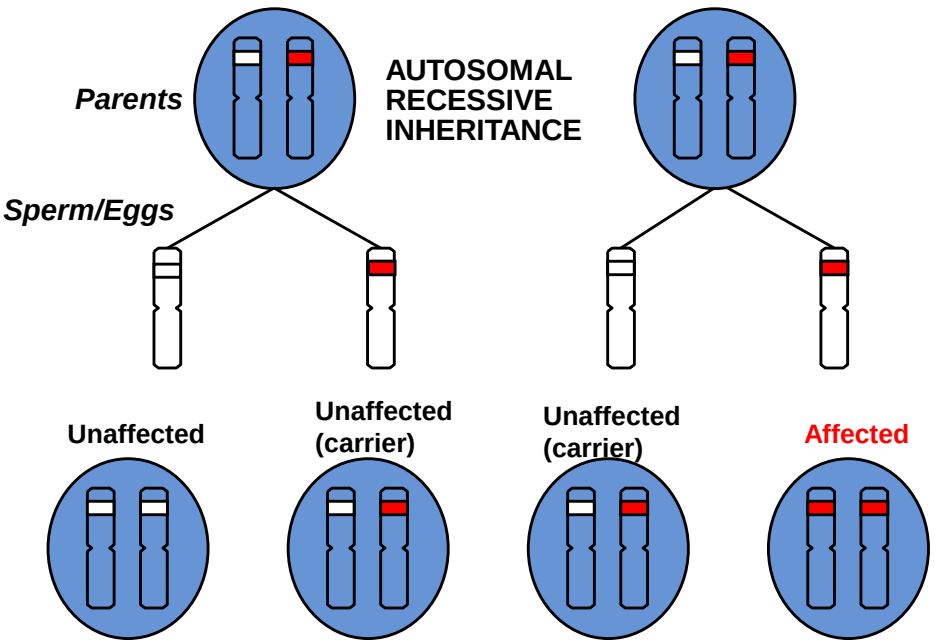
Parent who are carriers for the same autosomal recessive condition have one copy of the usual form of the gene and one copy of an changed gene of the particular pair



A parent who is a carrier passes on either the usual gene

or the changed gene into the eggs or sperm

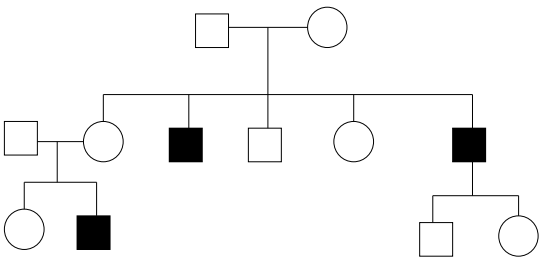
The other parent who is also a carrier for the same condition passes on either the usual gene or the changed gene into his/her eggs or sperm



Autosomal recessive conditions

- Haemochromatosis
- Sickle cell disease
- Thalassaemia
- Fanconi anaemia
- Spinal muscular atrophy
- Mucopolysaccharidosis type I (Hurler syndrome), other MPS except type II
- Many many more!

X-linked Recessive Inheritance

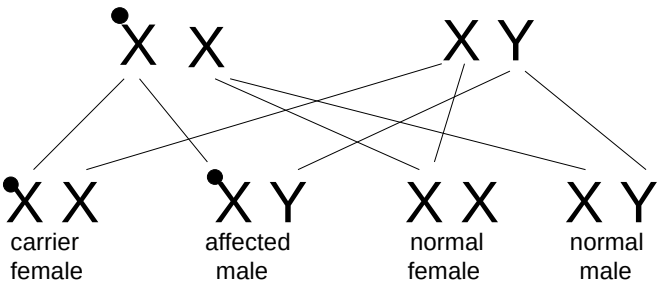


Examples:

- Duchenne/Becker muscular dystrophy
- Haemophilia
- Red/green colour blindness

- Usually only males affected
- No male to male transmission
- Females occasionally manifest due to pattern of X inactivation or with X chromosome abnormalities

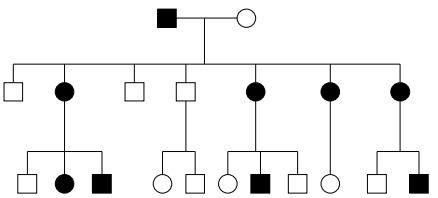
Segregation in X-Linked Recessive Disorders



- Half of sons of a carrier mother are affected
- Half of her daughters are carriers
- Daughters of affected males are obligate carriers
- Sons of affected males are always normal

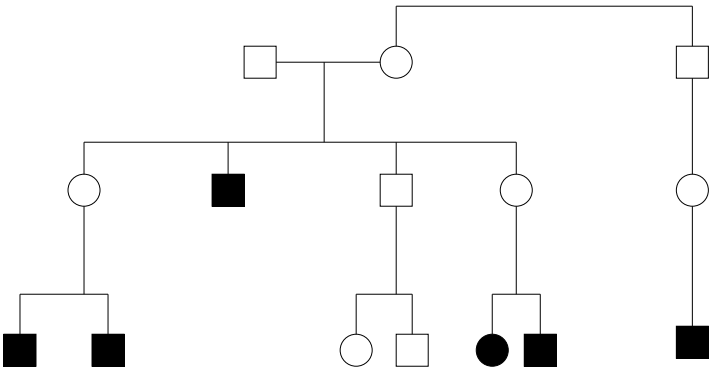
X-linked dominant

- Males and females affected
- Females usually less severely affected than males
- 1 in 2 risk to children of affected female
- All daughters of affected male affected
- No male to male transmission



If male lethality affected female produces offspring in 1:1:1 ratio normal female:affected female: normal male

Fragile X syndrome



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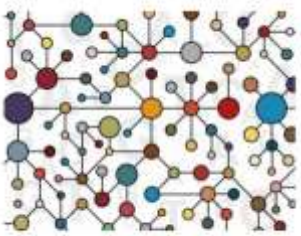
Online courses



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Thank you

Questions ?