

Women with 1 = carrier

Genetic diagram: show chromosome (X or Y) & allele (letter identifying normal/faulty) on X chromosome

19. Most phenotypic features result from multiple genes rather than single gene inheritance

20. Causes of variation that influence phenotype

genetic variation nature

Mutation: change to DNA base sequence – results in allele (different form of gene) – occur during cell division

Caused by organisms having different alleles – lead to differences in phenotype

Sexual reproduction causes genetic variation – results in alleles being combined in lots of different ways in offspring

environmental variation nurture: characteristics caused by organism's environment – acquired

21. Human Genome Project

Big idea: find every single human gene

Now they've all been found – scientists trying to find out what they do

Helped identify 1800 genes related to disease – high potential benefits for medicine

potential applications within medicine

Understanding genome can indicate persons risk of developing diseases

Sickle cell anaemia: changed shape of haemoglobin – but malaria can't hold to it (positive mutation)

Can help identify which medicines are best to treat illnesses – alleles can effect how medicines work on body

Gene therapy – cystic fibrosis

Prediction/Prevention of disease: different genes & lifestyle factors cause many common diseases (cancers / heart disease)

If doctors knew what genes made people likely to get disease, they could:

tailor advice to avoid it

regularly check us to ensure early treatment if susceptible disease is developed

Testing/Treatment for inherited disorders – caused by presence of faulty alleles in person's genome

Doctors can identify genes/alleles suspected to cause inherited disease faster – can test people for it – possible to develop better treatment/cure

New / better medicines

HGP highlighted common genetic variations – some affect how individuals react to certain diseases/treatments

Scientists can: design drugs tailored to people with particular genetic variation

determine how well existing drug will work for individual

Allow us to design more effective treatments with fewer side-effects

identify how some patients will respond to particular drug

identify appropriate dosage for patient

Drawbacks

Increased stress: told about potential of disease from young age

Gene-ism: people with genetic problems under pressure not to have children

Discrimination by employers/insurers: life insurance less likely for people with genetic likelihood of serious disease

22. There's usually extensive genetic variation within population of species through mutations

23. Most genetic mutations have no effect on phenotype

some mutations have small effect on phenotype

rarely, a single mutation will significantly affect phenotype