#MCBG 2032

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Phenylketonuria

single mutation can affect brain development however if the correct diet is followed then all detrimental health consequences can be ignored.

diabetes type II

lifestyle disease, the genetic component is estimated to change the likelihood of developing the disease by 2%, but the main causative factors of the disease relate to diet.

Lactose intolerance.

Individuals with lactose intolerance cant digest lactose, however the bacteria in their gut van which leads to the build up of gas. this can be very painful for the person in question.In humans lactose intolerance would naturally occur after weening however as cow milk is a staple part of most human diabetes the genes controlling lactase production are never switched off and the ability to digest lactose is not normally lost.

Himalayan Phenotype.

Cancer

In most cases cancer is not inherited. In breast cancer/ovarian cancer BS1, BS2 play some role in genetic disposition, perhaps 5-10%

Cancer normally sets in during old age as cells must accumulate a series of a specific set of notations, (exactly which set of mutations accumulated lead to cancer is highly individual)

Environmental factors

1. UV
2. X rays
3. Alcohol
4. Overcooked food (amines, heterocyclic, aromatic hydrocarbons-meat. acrylamide- potatoes
5. Azo dyes

6. tartrazine (food colourants)
7. Nitrate cured foods
8. Pesticides

Cancer initiators

agents which cause DNA damage (mutagens). these factors may be chemical, biological (such as HPV- human papilloma virus cause latent genetic damage- Cancer predisposition.

Cancer Promoters

Promotes excessive proliferation (does not directly damage DNA).

Examples

1. wounding
2. phorbol esters
3. HRT/ oestrogen (breast cancer)
4. hepatitis B (promotes stomach cancer)
5. HIV (Kaposi sarcoma, this disease is always present but only manifests itself in immunocompromised individuals)

Penetrance

percentage of the population who demonstrate at least some degree fo phenotypic expression.

Expressivity

reflects the range of expressions of the gene/allele present in the population.

Genetic complementation

two different mutations in heterozygous condition affecting the same protein/ pathway can compliment each other to cause a novel phenotypic effect.

examples

rare form of albinism: both mother and father were normally pigmented and there families had minimal instances of albinism. however both children where albino' s . Analysis found that this albinism stemmed not from those genes and mutation most commonly associated with albinism but rather from a combination of recessives (heterozygous) from both father and mother (mutations in TYRP1

gene), which on their own would have little or no effect on the pigment production pathway, but when combined reduced pigment production significantly.

NOTE: Rufus albinism leads to a phenotype with reddish hair, lighter skin, and blue grey eyes.

Forked line probability method.

##chromosome level (revise mitosis and meosis) copy slides chromosome basic structure #### chromosome groups

meta-centric

centromere is half way up the length of the chromosome

submetacentric

centromere is more to the one side of the chromosome than the other,

Acrocentric

the centromere is very far to the one side of the chromosome, with a long arm containing most of the genes and a short arm containing predominantly temolmeric DNA.

Telocentric

humans do not posses any telocentric chromosomes, but certain insect of crustation species do.

Holocentric

Centromere like structure exist along the entire length of the chromosome. this may decrease the chances of faulty division/ segregation.

chromosome banding

bands were named and used to locate specific genes. the banding patterns are due to uneven DNA densities in the coiled structure of the chromosome.

Size and shape of different chromosomes.

Chromosome level mutations

Anuploidy (Spelling)

anupliody refers to monoploidy or trisomy

Structural rearrangements

1. deletion
2. duplication
3. translocation
4. inversion
5. fission
6. fusion

Trisomy

when an individual inherits three copies of a particular chromosome. In Humans only three of all the possible trisomies are viable, (as in individuals with these mutations will still br born and not terminated during pregnancy)

of these three (Trisomy 13, 18, and 21) only individuals with trisomy 21 can survive past the first few years of childhood. Individuals with Trisomy 21 have down syndrome.

NOTE: the smaller relative size of chromosome 21, meaning that it contains less important genes may be related to its increased viability .

Monosomy

when an individual inherits only one copy of a particular chromosome.

Nullisomy

having no copies of a particular chromosome.

non disjuncture

(review mitosis and meiosis) If non disjuncture happens in meiosis it is generally worse than if it happens in mitosis as it will affect the whole organism.

Down Syndrome

occurs in about 1 of a thousand live births. originates from non disjunction in the egg cell rather than the sperm cell because the arrested development of the egg cells leads to decay of the separating and marking proteins as well as the

spindle fibre leaving the whole process open to more error. 8% of individuals survive for one year ?

turner Syndrome

monosomy X . occurs 1/20 000 births. ()only occurs in females webbed neck , short stator, underdeveloped ovaries lack of secondary sex characteristics.

Klienfelter syndrome

too many X's ,XXY , XXXY, XXXXY, XXXXXY. (XXYY) decrease in testosterone levels and as the more X's the higher the chance of brain damage.

XYY

XYY has the phenotype of a normal make. men with 2 Y chromosomes tend to be tall as there is a cumulative effect adding to height. (they may also exhibit increased aggression but this in unclear)

Polyploidy

most often flowering plants are polyploid, they are even specifically breed to have more chromosome sets as this usually increases fruit and flower size.

Examples

water melon must be bread from a tetraploid(?) and a diploid to get a tripliod infertile plant.

Kiwi fruit 12-16 copies

Strawberries 4 copies (tetraploid)

NOTE: plants are better adapted to polyploid because: 1. they are not as confined to a set physical form so different in growth and development genes operation levels are not so important 2. they can reproduce vegetatively so polyploid individuals aren't as severely evolutionarily disadvantaged.

Deletions

Piece of Chrome

sometimes multiple genes are lost when both strands break and a part of the chromosome is permanently lost. A specific deletion on chromosome 5. (where the entire p arm is deleted leads to a serious syndrome)

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14/02/2018

acids and bases.

pH

$pH = -log_{10}[H^(+)]$

####ka

$k_a = \frac{[A^-][H^+]}{[HA]}$

pKa

$pka = \log_{10}(pk_a)$

#APES

Survivor ship curves

insert appropriate graphs.

Type1 1

Typical populations

domestic pets and humans in the developed world

Type 2

Typical populations

1. seeds in a seed bank
2. marmites.

Type 3

typical populations

large mammals.

Reproductive Rate

combination of many factors. 1. length of reproductive season. 2. (reproduced output) number of offspring produced. 3. Investment 4. generation length (time between birth of mother and birth of daughter) 5. age of maturity.

potential population rate of change.

how fast populations can change. r_m *or* r are common symbols for the maximum growth rate.

K selecting

adaptions which favour ability to survive over the ability to reproduce. well adapted to predictable, crowded environments with significant resource scarcity.

general traits

1. large
2. can deffer reproduction to a later time.
3. iteroparous.
4. high investment per offspring
5. body resources invested more into survival than reproduction.

R selecting

unpredictable environments, periods/ areas of sudden abundance.

general traits

1. small(er)
2. earlier maturity
3. invest in reproduction versus survival
4. many offspring but with low investment per offspring.

Variation in abundance over time.

1. patchy resource allocation
2. inter-species interactions (predation, symbiosis)
3. environment
4. social structure

resources.

physicochemical. conditions.
1. temperature 2. humidity 3. occupancy.

defining environments.

environments are a smooth continuum of suitable (good) to unsuitable (bad). There is a threshold below which conditions are too ad to sustain a particular organism, below this line there are no available habitats. above this line there is spectrum of habitats varying from poor to good, with associated increase in survival and reproductive rates. ##### Examples Elephants in the Kruger

because of artificial water easy for herds with young elephants to move around which otherwise they would be unable to migrate. artificial water sources. this artificially increased the carrying capacity.