#MCBG 2032

13/02/2108

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

basics

A cancer is a group of cells whose proliferation is uncontrolled, and which can spread to other locations in the body which are normally populated bu other cell types.

benigin tumours

grow but do not spread.

malignant tumours

over proliferate and invade other body tissues/ areas in the body.

types of cancers

Carcinomas

derived form epithelial cells

Sarcomas

derived from connective or muscle tissues. Known as Osteosarcoma in the case of bone tissue and Kaposi sarcoma in the case of soft connective tissue.

Lymphoma and leukaemias

1. Cancers of the hematopoietic system.
2. Lymphoma (solid tumour)
3. Leukaemia excess of circulating immature blood cell precursors. ##### Cancer of the nervous system brain and central nervous system.

NOTE: most common in children

Germ-line cancer.

Cancers of breast, prostate, lung, pancreas, and colon.

Causes of cancer.

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

hereditary.

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 (hetercyclic amines,polycyclic 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)

stages of cancer development

1. normal cells (mutation occcurs)
2. increased ability to proliferate (mutation)
3. 1-2mm tumour of rapidly proliferating cells which do not undergo apoptosis of differentiation. (mutation)
4. vascularized growing tumour (mutation)
5. large tumour capable of invading near by tissue (metastasis)

NOTE: cancer results from a series of somatic mutations, affecting the same cell, and different cancers are genetically heterogeneous.

Genetic instability in cancers.

deficient local DNA repair leads to the accumulation of point mutations increased chromosomal instability and gross genome abnormalities.

summary of key properties

1. disregards ex/in growth regulation signals.
2. avoid apoptosis, differentiation and replicative senescence
3. genetically unstable.

4. invasive
5. metastatic (survive and proliferate in foreign) sites.

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

Aneuploidy (Spelling)

Each cell is has at least one extra chromosome or is missing at least one chromosome

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 be 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.

Down syndrome (Trisomy 21)

Frequency: 1/1000 Effects 1. mental retardation 2. short stature 3. heart disease 4. shortened life span.

Edward Syndrome (Trisomy 18)

Effects: 1. kidney and intestinal malformation 3. heart defects. 4. mental retardation

NOTE: only 8% survival past the first year.

Patua syndrome (Trisomy 13)

Effects: 1. Kidney and hear defects 2. polidactily (too many digits) 3. Nervous system abnormalities 4. Death within the 1st year.

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.

Polyploidy

Structural rearrangements

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

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

Frequency 1/20 000 (only occurs in females) Effects: 1. webbed neck 2. short stature 3. underdeveloped ovaries and lack of secondary sex characteristics.

Klinefelter syndrome XXY

Too many X's ,XXY , XXXY, XXXXY, XXXXXY. (XXYY) Effects: 1. Decrease in testosterone levels 2. 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)

XXX

Frequency: 1/1000 Effects: 1. Normal female with normal fertility.

NOTE: this conditions is seldom diagnosed.

Polyploidy

Three or more complete sets of chromosomes present in somatic cells. #### 2n-20n caused by non reduced gamete fomration, which is very rare in males and non viable.

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)

Frogs *Xenopis levis* 4n *Xenopus tropicalis* 2n

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. 3. less precise sperm targeting is necessary.

Deletions

deletions often occur by chromosome breakage during the cell cycle. These deletions may be terminal of interstitial. If the centromere is lost, the entire chromosome will be lost.

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)

Cri du chat (5p-)

Frequency: 1/20000-50000 Effects: 1. Intellectual disability, 2. low muscle tone 3. microcephaly 4. distinctive facial features. 5. normal life expectancy

Duplications

Cause: retro transposition, or non-allelic homologous recombination in repeat rich areas.

A small region of a chromosome is repeated, and placed next to the existing copy (in tandem). duplications tend to happen in high repetitive sequences of DNA. These repeated regions mean that the chromosome will note line up very well with its homologous pair.

passed on by unequal order, and may or may not have a phenotypic effect.

Huntington Purea

Repeats alter gene function, which can lead to brain damage. Repeats may have a positive effect because they allow one copy of a gene to evolve independently, specialise in a different function while the original function is still conserved by the other copy of the gene.

Inversions.

Cause: non-allelic homologous recombination Effects: 1. pairing in meiosis occurs via looping 2. cross over in the looped region can cause major deletions or duplications.

Translocation

Effects: 1. novel chromosomes are generated 2. individuals are usually infertile. 3. can result in aneuploidy (such as 14q21q inherited down syndrome) 4.

terminal

A piece breaks off a chromosome and sticks to the end of a non-homologous chromosome.

reciprocal

Exchange of pieces between non-homologous chromosomes #### Robertsonian

two Acrocentric chromosomes lose their short arms and get stuck together.

Fission

deletion (loss of acentric piece)'

Fusion

bicentric chromosomes (breakage in mitosis)

breakage fusion bridge cycles in cancer.

Copy image. 1. end of a chromosome breaks off (during meiosis) 2. one daughter cell inherits the chromosome lacking a telomere. 3. the new cell enters S phase and replicates its DNA (it now has two sticky DNA ends right next to each other.) 4. loose DNA strands bind to form continuous DNA loop. 5. fused sister chromatids are pulled apart during mitosis, causing breakage at a new sight 6. one daughter cell inherits chromosome with duplicated genes and again a free sticky end. this cycle can then begin again leading to even further chromosome distortion.

Imprinting

Hinny vs mule

Mule

horse mother donkey father hardy and obedient

Hinny

donkey mother, horse father. temperamental, untrainable.

Intersistial deletion of chromosome 15

Prader -Willi syndrome

Cause: deletion from father, ZNF127 and IPW inactivated on the maternal copy. effects: 1. mental retardation 2. obesity 3. diminished growth.

Angelman syndrome

1. deletion from mother: UBE3A inactivated on paternal copy.

2. mental retardation
3. epilepsy
4. lack of motor development.

X inactivation

the inactivation of one of two X chromosomes in females via DNA methylation.

NOTE: which X chromosome is inactivated varies randomly from cell to cell.this inactivation process turns the inactive X into a bar body, (highly methylated and rolled up in histone proteins).

MCBG 2036

14/02/2018

acids and bases.

pH

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

####ka

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

pKa

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

dependant and independant variables.

Buffers

NOTE: buffers may be temperature sensitive, for example Tris buffers. phosphate buffers are more reliable but it is still best to take temperature into consideration

Indicator dyes.

examples

1. phenlythyelian (spelling)
2. bromothymyl blue (spelling)
3. methyl orange

paper strips

inaccurate but useful for rough estimations of pH or to identify changes in pH

Electrode

accurate to two decimal places.

#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.

#CHEM 2003

Valence Bond Theory

History

Valence bond theory was the First quantum mechanical theory developed. It was developed by Walter Heitler, friz London and Linus Pauling, and helped to explain concepts which were poorly predicted by VSPER theory such as diamagnetic/ paramagnetic properties.

Basics

Wave functions.

the wave function of an electron pair is formed by superimposing the wave function for the separate fragments of a molecule.

Electrons

Each electron will have a wave function ψ that describes the behaviour of that electron. (section 1.2 in book)

the probability of finding an electron at any given location is given by $\chi = \psi^2$

$/chi$ is used to represent the orbital of a given electron.

bonds

each bond will also have a wave function which describes the movement of electrons within that orbital.

examples

the bond between two hydrogen can be expressed as $\psi = \chi_A(1)\chi_B(2)$ (or $\psi = \chi_A(2)\chi_B(1)$ is just as valid)