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GENETICS LECTURE NOTES 2014:

Genetic disease = are those diseases which are inherited from the parents and can be transmitted to the next generation.

Genetic counseling is the process by which individuals or relatives are at risk for disorder that may be hereditary are advised of the consequences of the disorder, the probability of transmitting it and ways of in which this may be prevented, avoided or ameliorated.

COMMON CHROMOSOMAL DISEASES

1. Down Syndrome

- Chromosomal abnormality,
- Trisomy 21
- Usually present at birth
- **Clinical features** (small low set ears, up-slanting eyes, prominent epicanthic folds, flat facial profile, protruding tongue, short neck, mild short stature, short broad hands)

Associated conditions:

- Congenital heart disease e.g. VSD < ASD, fallot's tetralogy
- Deafness
- Dementia
- Cataract

Investigation: chromosomal analysis showing additional chromosome 21

Management: Patient needs cardiac investigations and testing for hearing.

1. Klinefelter's syndrome

- Boys with klinefelter syndrome enter puberty normally but by mid puberty the testes become small
- Infertility due to azoospermia, Gynaecomastia. They have decreased testosterone.

Investigation: Chromosomal analysis.

Test to prove the azospermia is testosterone.

1. **Patau syndrome**

- Trisomy 13
- Microcephaly, Microphthalmia
- Cleft lip and palate

Investigation: chromosomal analysis

1. **Edwards Syndrome:**

- Trisomy 18
- Features: Congenital heart disease
- Low birth weight
- Overriding fingers
- Usually they die within 4 days. Rarely they can live several months.

1. **Turner's syndrome**

- Most girls have single chromosomal (45, XO)
- Features: Short stature, broad neck, ptosis, widely spaced nipples, congenital heart disease,

GENETIC DISORDERS WITH CARDIAC FEATURES

1. **Marfan syndrome**

- Autosomal dominant multisystem disorders caused by mutation in the in the chromosome 15.
- Features: Include, tall and slim body build with long limbs scoliosis, long fingers, aortic aneurysm

1. **Di George syndrome**

- Short stature
- Congenital heart abnormality e.g. tetralogy fallot
- Prominent nasal bridge

1. **Williams syndrome**

- Autosomal dominant
- Peri-orbital fullness, short stature, congenital heart abnormality chest deformity.

GENETIC DISORDERS WITH LEARNING DISABILITY

1. **Fragile X syndrome**

- It is commonest inherited disease of mental retardation
- Boys with Fragile X syndrome usually have global developmental delay
- Can affect both girls and boys
- They boys have stereotyped repetitive behavior such as hand flapping and resistance to change of routine.

1. **Prader-Willi Syndrome**

- Babies are frail and may fail to thrive
- Older children have learning difficulty and short stature.

Genetic disorders with neuromuscular features:

1. **Congenital Myotonic Dystrophy**

- It is an autosomal dominant with onset usually in adult life
- At birth the baby is floppy
- During pregnancy there is polyhydramnios

- Usually the affected children are from women who are also have myotonic dystrophy

1. **Duchene muscular dystrophy**

- Presents with development delay
- Child climb up his thigh when standing up (Gower's sign)
- Diagnosis is by genetic testing
- X-linked recessive
- Mean age of onset is 5 years.

1. **Spinal muscular atrophy**

- Autosomal recessive disorders
- Symmetrical proximal weakness as a consequence of degeneration of anterior horn cells of spinal cord

- Investigation: genetic molecular testing

GENETIC DISORDERS WITH DERMATOLOGICAL FEATURES

1. **Ehlers Danlos syndrome**

- Autosomal dominant
- Hypermobility of small and large joint with soft skin
- Soft skin which hyper-extensive

1. **Neurofibromatosis**

- NF1 has a autosomal dominant disease
- Café au lait spots
- Neurofibroma
- Chance of transmitting it to children is 1:2

1. **X-linked hypohydrotic ectodermal dysplasia**

- It is X linked recessive
- Boys have reduced sweating which may cause dangerous hyperpyrexia in infancy

1. **Tuberous sclerosis**

- Autosomal dominant
- Characterized by hamartoma's on the skin, brain and other organs
- Commonly presents with infantile spasm, seizures and mental retardation

OTHER INHERITED DISEASES

1. **Polycystic kidney disease:**

An autosomal dominant disease is associated with vascular abnormality like berry aneurysm. The kidney is usually palpable bilaterally and patient has hypertension.

1. **Cystic fibrosis:**

This is an autosomal recessive disease: meaning there is 1:4 chance of transmitting to another child. Child presents with failure to thrive and pancreatic insufficiency and recurrent chest infection.

Investigation: sweat test

1. **Glycogen storage disease:**

Specific enzyme defects preventing mobilization of glucose from glycogen and resulting in abnormal storage in liver and muscle. Patient usually presents with hypoglycemia in the morning.

This is autosomal recessive condition usually presenting with hypoglycemia, lactic acidosis, poor growth, mental retardation.

1. **Haemophilia:**

This is a X-linked recessive disease affecting male children and presents with bleeding into the muscles and joints.

1. Achondrioplasia:

Short limbs, autosomal dominant lumbar lordosis, large head.

1. Von Willebrand's:

An autosomal dominant disease causes high bleeding time and bruises and bleeding.

POLYGENIC INHERITANCE DISEASE:

These are disease which can run in the family but there needs to be interaction with external environment which include the following:

- A. Diabetes
- B. Multiple sclerosis

AUTOSOMAL RECESSIVE DISEASES:

- 1. Sickle Cell Disease
- 2. Thalassemia
- 3. Cystic Fibrosis

AUTOSOMAL DOMINANT DISEASES:

- 1. Polycystic Kidney Disease
- 2. Huntington Disease
- 3. Neurofibromatosis
- 4. Von Willibrand Disease

X-LINKED DISEASES:

- 1. Duchene Muscular Dystrophy
- 2. Haemophilia

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