تجمع الرياض الصحي الثاني
Riyadh Second Health Cluster

Aneuploidy

Aneuploidy

It is the second major category of chromosome abnormalities in which chromosome number is abnormal in the cells such as Down syndrome 47, XY, +21.

Referral categories

- Family history of structural or numerical abnormalities similar or related problems might suggest mode of inheritance
- Congenital malformations of the fetus during pregnancy that phenomenon through ultrasound.
- Mother's age, health or other exposures in pregnancy such as some medications or exposure to certain types of X-ray.
- Dysmorphic facial features
- Parents who carry the genetic abnormalities

Sample requirement

- 5ml Blood sample in Lithium heparin tube and for Neonatal referrals (< 1 month) – 2ml
- Also request EDTA specimen if Molecular Genetics studies are required for more investigation.

Common Aneuploidy syndrome

Down syndrome:

Down syndrome (DS) result from extra copy of chromosome 21 that means each cell in the body has three copies of chromosome 21 instead of the usual two copies.

However, less common Down syndrome can be occurring as a result of a structural abnormalities between chromosome 21 and any of others

chromosomes (nondisjunction translocate) during crossing over (eggs and sperm).

Rarely, extra copy of chromosome 21can be occur in some body cells, this condition is called mosaic Down syndrome. However, parents could pass these genetic abnormalities when one of them is carrier. Carrier parent's means one of them have balanced rearrangement between chromosome 21 and other chromosome that is means no gain or loss for genetic material.

It is a test that evaluates the number and structure of chromosome, to diagnose genetic diseases, some birth defects, and certain disorders of the blood and lymphatic system.

Down syndrome is a chromosomal condition that is associated with a variety malformation such as heart defect, characteristic distortion facial feature, mild to

moderate mental retardation and muscle weakness in childhood. All downs' patient suffers from cognitive delays digestive abnormalities. DS consider high risk of childhood leukemia and other diseases such as Alzheimer. The possibility of Incidence 1 in 800 live births 90-95% cases has numerical abnormalities of chromosome 21, 85% comes from mother contributes and 15% from the father. One the other hand 3-4% resulting from robertsonian translocations and ~ 1% Mosaic.

Clinical features of DS

- Dysmorphic features.
- Hypotonia.
- Physical, psychomotor and mental development are affected.
- IQ ~ 30-70.

- Short neck.
- Small head.
- Upward slanting eye lids (palpebral fissures).
- Unusually shaped or small ears.

Risk factors:

- Advancing maternal age.
- Carrier parents of the genetic translocation for Down syndrome (robertsonin)
- Having had one child with Down syndrome.

Diagnosis tests:

1) Prenatal Diagnosis tests:

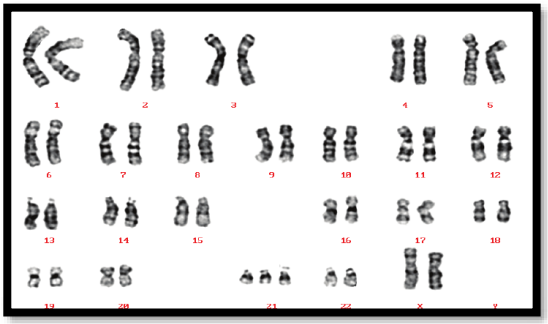
- **Ultrasound:** typical structural abnormalities may be detected on prenatal ultrasound and

may raise the suspicion of aneuploidy syndromes

- **Amniocentesis** at 15-20 weeks
- **Chorionic villus sampling** (CVS) At 8–12 weeks

2) Postnatal diagnosis tests:

- **Chromosomal analysis (G-band karyotype)**



ISCN: 47,XX,+21

Chromosomal microarrays

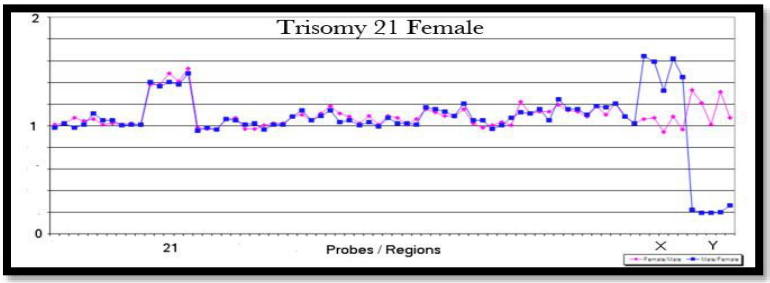


Figure showed Down syndrome demonstrated by microarray technique.

Fluorescent in Situ Hybridization (FISH)

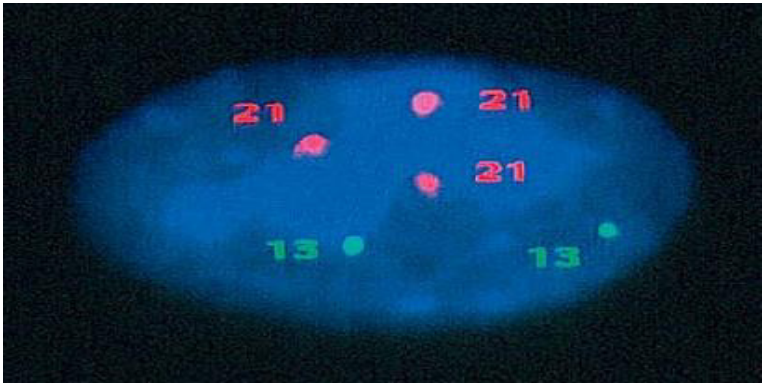


Figure showed Down syndrome demonstrated by Fluorescent in Situ Hybridization (FISH).

Down Syndrome Treatments

Down syndrome has no specific treatment as is the case with many genetic diseases. However, Early treatment physical and developmental therapies programs can help can help down patients to reach their full potential. Also, may include speech, and specific educational therapy.

لأن الوعي وقاية ..

إدارة التثقيف الصحي

Clinical Pathology Department

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