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Fanconi Anemia

POSTED BY: MEDICAL PPT

What is Fanconi Anemia?

Fanconi Anemia is an autosomal recessive disorder.

We all have FA genes.

Known mutations involved: FANCA, FANCB, FANCC, FANCD1, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCI, FANCI, FANCI, and FANCM

Fanconi Anemia predisposes an individual to numerous types of **cancer**, including **acute myeloid leukemia**, **breast cancer**, **squamous cell carcinoma** of the head and neck and cancers of the gynecological system, **skin**, esophagus, liver, and kidney

Common **presentations** of patients with Fanconi Anemia:
Anemia

Pancytopenia

Bone marrow hypoplasia

Thrombocytopenia

Low Birth Weight

Short stature

Absence of or malformity in hands and arms, for example the absence of a thumb or the presence of polydactyly

Presence of only one kidney or of a horseshoe kidney

Gene therapy to target this role of FA genes in individuals with FA gene mutations.

Diagnosis

DEB and MMC tests

Diagnosis typically occurs before the age of twelve (Fanconi Anemia Research Fund, Inc., 2006).

MMC test is **used** to diagnose Fanconi Anemia at the University of Kentucky.

Subtyping via use of retroviruses needs to be incorporated into standard protocol when diagnosing a patient with Fanconi Anemia.

Treatment

Retrovirus mediated gene transfer

Lentivirus mediated gene transfer

Risks of Gene Therapy

Retrovirus potential to stimulate oncogenes.

Lentivirus association with arthritis and encephalitis in goats, leukemia in cattle, anemia in horses, and immunodeficiency

Fanconi Anemia ppt

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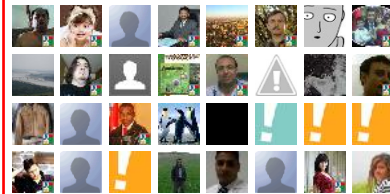
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