Fanconi Anemia

POSTED BYE-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, FANCJ, FANCL, 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 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

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