Fanconi Anemia

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Genomics and Medicine

Pictures courtesy of http://www.fanconi.org/
General Overview

- Around 500 families
  - ~1800 cases in medical literature
- Usually symptoms appear from birth
- 13 genes involved with FA
- Life expectancy – 20 to 30 years

Symptoms include:
- Bone marrow failure
- Physical deformities
- Predisposition to many cancers

Pictures courtesy of http://www.emedicine.com/PED/topic3022.htm
Classical Diagnosis

- Looks for physical symptoms:
  - Short stature
  - Skin discoloration
  - Hand and arm abnormalities
  - Fatigue
- Definitive test:
  - Chromosome breakage test
Classical Treatment

• Palliative Treatments
  – Frequent blood count checks/Blood transfusions
  – Bone marrow transplant
  – Hormone therapy to stimulate RBC growth
  – Chemotherapy for the cancers
Genetic Diagnosis

- FA is related to 13 genes.
  - FANCA, FANCB, FANCC, FANCD1, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCIJ, FANCL, FANCM, FANCN
  - If any one is homozygous recessive, FA will present itself.
  - FANCA -16q24.3, FANCC -9q96.9
  - DNA tests can also identify specific mutations in Fanconi genes.
Genetic Treatments

• Ongoing clinical trials by National Institutes of Health Clinical Center. Sponsored by NHLBI

• Gene Therapy
  – replace the diseased genes in the stem cells of bone marrow with healthy genes, then destroy remaining diseased cells.

  – Earlier definitive testing
References

• Online Mendelian Inheritance in Man
• Fanconi Anemia Research Fund, Inc.
• MedlinePlus Medical Encyclopedia
• Clinicaltrials.gov
• NCBI Pubmed