Fanconi Anemia

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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

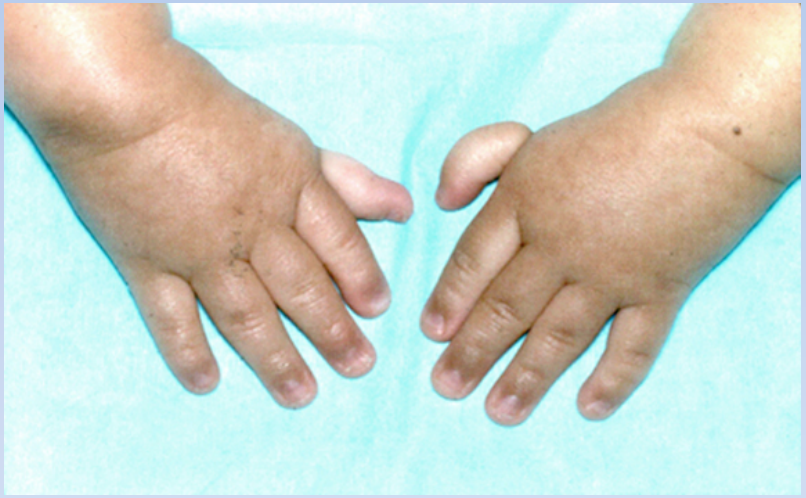
Symptoms include:

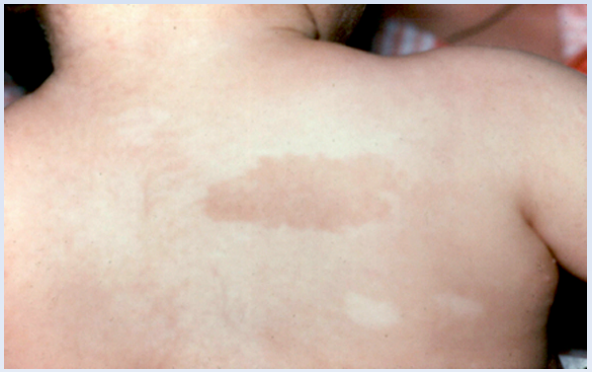
- Bone marrow failure
- Physical deformities
- Predisposition to many cancers

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

Classical Diagnosis

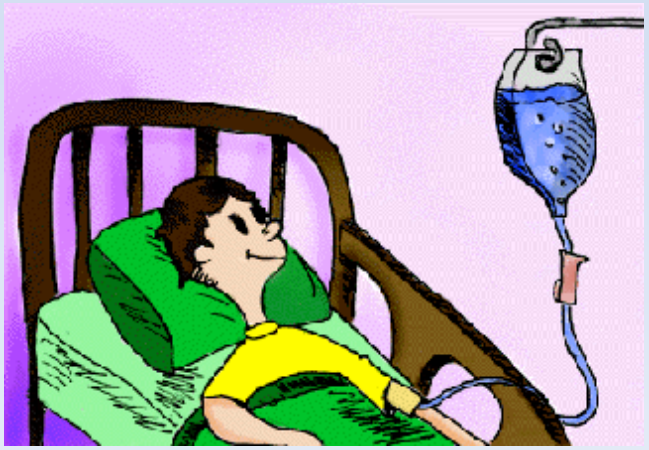
- Looks for physical symptoms:
  - Short stature
  - Skin discoloration
  - Hand and arm abnormalities
  - Fatigue
  - Definitive test:
    - Chromosome breakage test





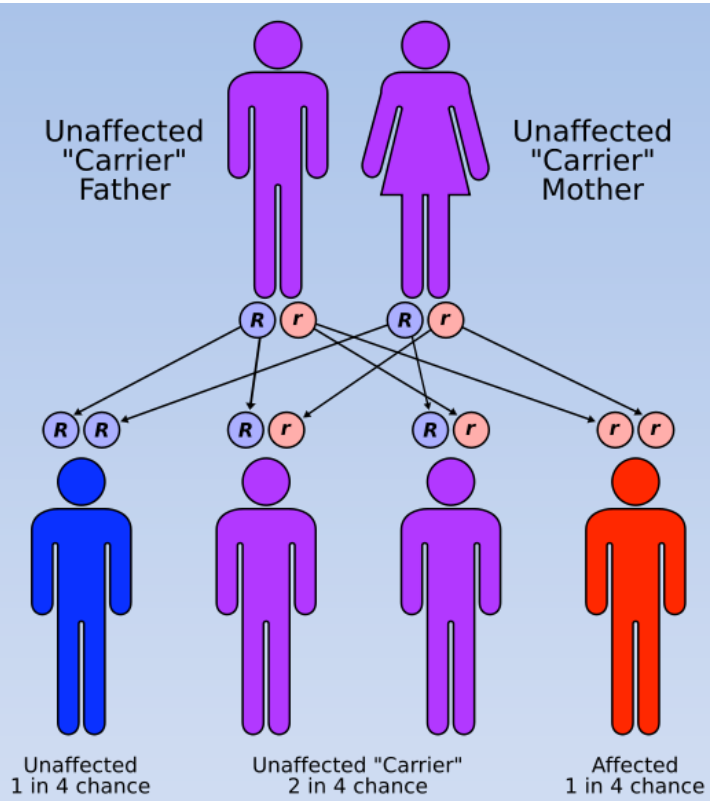
Classical Treatment

- 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, FANCJ, 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