

## **Condition:** Fanconi anemia

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### **Inheritance:**

Autosomal recessive.

### **Genetic etiology:**

Genetically heterogeneous; mutation in various genes involved in DNA repair.

### **Frequency:**

Approximately 1/100,000.

### **Clinical features:**

Common features include short stature, skin hyperpigmentation with café-au-lait spots, malformations of the upper limbs (absent, hypoplastic, or supernumerary thumbs), ocular anomalies (microphthalmia, strabismus), renal anomalies (horseshoe or dysplastic kidneys), hypogonadism, and gastrointestinal anomalies. There is a progressive bone marrow failure leading to pancytopenia and a greatly increased risk of cancer.

### **Management:**

Avoidance of sun exposure; supportive care.

### **Genetic counseling:**

Parents of affected child have 25% recurrence risk; molecular genetic testing for some of the genes responsible; diagnostic testing most often done by testing for chromosome breakage upon exposure of cultured peripheral blood cells to diepoxybutane.