

Cancer Prone Disease Section

Short Communication

Diamond-Blackfan anemia (DBA)

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Identity

Inheritance

Genetic heterogeneity; majority of cases autosomal dominant, occasionally with variable expression (incomplete dominance) manifesting as mild anemia in transmitting parent; some cases apparently autosomal recessive, not linked to 19q.

Clinics

Phenotype and clinics

- Chronic constitutional are generative anemia with absent or decreased red cell precursors in bone marrow;
- Macrocytosis, elevated fetal hemoglobin, increased red cell adenosine deaminase;
- Physical abnormalities in 30% of DBA cases including craniofacial and thumb abnormalities, atrial or ventricular septal defects, short stature, mild retardation, etc.

Neoplastic risk

- Hematologic malignancy: in 2.5% of all reported cases of DBA; primarily ANLL with no FAB preference but also ALL, Hodgkin's disease;
- Solid tumors include carcinoma of liver, stomach, osteogenic sarcoma;
- Age of malignancy onset from 2 to 43 years;
- Disease-related and treatment-related factors, i.e., allosensitization and iron overload, contribute to malignancy.

Treatment

Corticosteroids, transfusion, bone marrow transplant.

Evolution

Some patients enter remission, with or without corticosteroid therapy.

Prognosis

Median survival: 38 years.

Genes involved and proteins

RPS19

Location

19q13.2

Protein

Description: Ribosomal protein S19; ribosomal proteins are a major component of cellular proteins; their function(s), aside from being part of the ribosome, are unknown.

Mutations

Germinal: Mutations in RPS 19 identified in DBA patients include non-sense, frameshift, splice site and missense mutations; three patients had disease-associated chromosomal abnormalities in DBA region, t(X;19), t(8;19), microdeletion 19q.

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