Qure
Center for Complete Care
Blood Diseases. Cancer. Stem Cell Transplantation

Diamond-Blackfan anemia (DBA)

Diamond-Blackfan anemia (DBA) is a condition in which the child has a low red cell count (anemia) due to a genetic problem that affects the ability of the body to make red blood cells.

What causes DBA?

DBA is an inherited disease affecting the ability of special cells in the bone marrow to make healthy red blood cells (bone marrow failure syndrome). The most common gene defect involves ***RPS19***, which affects the protein-manufacturing ability of those cells, but other gene defects have been found, and in a minority of patients no gene defect is identified.

How is DBA diagnosed?

DBA presents most commonly in the first year of life with increasing pallor (pale skin) due to anemia. Over one-third of patients also have congenital abnormalities, affecting the face, forearm, thumbs, kidneys, heart, and eyes.

The **complete blood count (CBC)** is a simple blood test that can be done in almost any medical lab. In DBA this shows severe anemia, with larger red cells, and fewer young red cells **(reticulocytopenia)**, while the rest of the blood cells such as white cells and platelet cells are usually normal. Along with the physical appearance, this may be diagnostic, but a **bone marrow aspiration** is often done to confirm a lack of red cells.

Other special blood tests for confirmation include:

- red cell enzyme (adenosine deaminase)
- fetal hemoglobin
- gene testing

How is DBA treated?

- **Corticosteroids (e.g. prednisone)** is usually effective and the goal is to give a trial of an adequate dose for a month then gradually reduce to a low dose to avoid side effects which include fractures, cataracts, growth retardation, high BP and diabetes. Due to these side effects, steroids are not given to young children (less than 6-12 months of age) for whom blood transfusions are initially given. Once steroids are started, around 40% of patients will continue at a low dose, 40% will not respond or need too high a dose and be switched back to blood transfusions, and 20% will go into remission and be able to stop all treatments.
- **Blood transfusions** are given to patients below 12 months of age, and those who do not respond or are needing too high a dose of steroid, to avoid the side effects of steroid. Chelation with deferasirox is essential with chronic transfusions to avoid organ (liver, heart) overload with iron.
- **Bone marrow transplantation** should be considered for patients who have an HLA-matched related donor, especially if are transfusion-dependent and under 5-10 years of age.

Can future children get DBA?

DBA is an inherited disorder, so there is a definite risk, which may be as high as 50%. Professional genetic counselling is essential because the risk of recurrence will depend on whether either of the parents have the same gene defect or evidence of DBA. If a mutation is identified in the family (e.g. *RPS19*) *prenatal diagnosis* is possible as well as *preimplantation genetic diagnosis* (PGD) to reduce the risk of another affected child, your counsellor will discuss these options with you in more detail.

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How often do I have to see the haematologist?

Typically a history and physical examination with blood count monitoring is done at 3–6 month intervals in patients who are stable. Patients who are weaning their steroid may need to be seen more frequently. Patients receiving blood transfusions should be monitored monthly with a clinic visit 3 monthly.

What are long-term outcomes for DBA?

With advances in supportive care, blood transfusion, steroid therapy, and BMT, patients are now surviving late into adulthood in US/Europe. There is a slightly increased risk for blood cancers and complete bone marrow failure so ongoing follow up is needed. Women with DBA who wish to become pregnant need to be followed at specialty centers as there is an increased risk to the fetus, and a possibility that the newborn might have DBA. Steroid-responsive women also often become transfusion-dependent during pregnancy.

Where can I get more information on DBA?

DBA is an extremely rare condition impacting only 100 new patients each year in India. Some recognized online resources from the U.S.A. which might be helpful:
www.dbafoundation.org
https://rarediseases.org/rare-diseases/anemia-blackfan-diamond