

Diamond Blackfan Anemia

Diamond Blackfan anemia (DBA) is a rare blood disorder in which the bone marrow does not make enough of the red blood cells that carry oxygen throughout the body. In addition to the anemia, several birth defects or abnormalities are often associated with DBA. The most common of these involve the head and face, arms and hands, heart, kidney, urinary tract, and genital organs. Many people with DBA also experience poor growth resulting in less-than-expected height. Because of the severe anemia that results from DBA, the condition is usually diagnosed within the first year of life. An estimated 25–30 new cases of DBA occur each year in the United States.

Once DBA is diagnosed, there are several treatment options, but the response to treatment varies among individuals. Corticosteroid therapy stimulates the production of more red blood cells in the majority of children treated, but it can result in serious side effects in the long term, such as growth retardation, diabetes, glaucoma, high blood pressure, and bone weakening. People who do not respond to steroids or who require too high a dose need routine blood transfusions to provide red cells. Frequent blood transfusions can cause excess build-up of iron, which, in turn, can cause damage to tissues and organs, particularly the heart and liver. Currently, the only cure for DBA is stem cell transplantation, which is limited to children younger than 16 years of age and requires a well-matched donor.

Not all people with DBA have the severe anemia that results in early diagnosis. People with milder forms of DBA often escape detection and may develop complications later in life. Because DBA is so rare, even severely affected people may have difficulty accessing state-of-the-art medical services, information, and support.

A Public Health Approach

Surveillance, Epidemiology, Research Clinical Resource Center Network

The Centers for Disease Control and Prevention (CDC) is working with a network of DBA clinical resource centers to advance research and improve the quality of life of people with DBA. At these centers, specialists from many fields work closely with local health care professionals to meet the individual needs of people with DBA. The clinical resource center network aims to:

- Provide specialized health services to help prevent and reduce complications of DBA.
- Learn whether the care received at these centers makes a difference in the lives of people with DBA.
- Increase awareness of DBA among health care professionals to improve early diagnosis of people with DBA.
- Gather important health information about people with DBA by encouraging them to enroll in the Diamond Blackfan Anemia Registry (DBAR). This information will help support research on the genetics, treatment, and the quality of life of people with DBA.

Public Health Surveillance Registry

CDC is providing funding to the clinical resource centers to develop a way to identify health care providers in the United States who are caring for people with DBA. CDC will encourage them to include their DBA patients in a surveillance system to monitor this care. This surveillance will include people with “classic” DBA characteristics – both those already participating in the DBAR and those that are not yet enrolled – as well as people with “nonclassical” disease. Given the discovery of several DBA-causing genes in recent years, family members with nonclassical presentations of the disease have been discovered.



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Studies of people with DBA suggest that some people with these DBA-causing genes do not have the severe anemia that usually accompanies the disease. Instead, these people have other diagnoses, such as birth defects or early-onset cancer, suggesting that the clinical spectrum of the disease is much broader than originally described. By including people with nonclassical DBA in this surveillance, there may be an opportunity to reach a better understanding of this disease. The knowledge gained through this surveillance program will help health care professionals make more accurate diagnoses, resulting in a better chance for appropriate treatment for their patients. Additionally, this information may be able to help other fields of care, including birth defects and pediatric cancer, which are common among people with DBA.



Informatics and Infrastructure

CDC supports data collection and analysis for research activities on DBA. Because each health care provider may care for a very limited number of people with DBA in his or her practice, it is necessary to gather data from many providers and settings so that enough data will be available to provide meaningful information.

It is important to design and offer data collection systems that are flexible and minimize the burden of data reporting. To meet this requirement, informatics systems provide the capability for investigators and collaborators to submit data in a variety of formats including paper forms, electronic submission to a website, and export of data from third-party software or information systems. Additionally, a sound informatics infrastructure can help coordinate and align data from multiple sources within a health care setting (e.g., clinical and laboratory data).

Health Education and Health Literacy

CDC, together with nurses and doctors from DBA resource centers, has developed a patient brochure and a series of fact sheets about DBA for families and others are in development. The brochure provides newly diagnosed people and their families with a general overview of DBA. The fact sheets focus on specific treatment options and provide more detailed information together with lists of other available resources, information, and support. The brochure and fact sheets are available online and include:

- Chelation Therapy fact sheet
- Corticosteroid Therapy fact sheet
- Stem Cell (Bone Marrow) Transplant fact sheet
- Blood Transfusion Therapy fact sheet
- Resources fact sheet
- Managing Your DBA Care: A Notebook for Families
- Congenital Anomalies fact sheet
- Parenting Corner Q & A fact sheet
- For Brothers and Sisters of Children with DBA fact sheet
- Genetics of DBA fact sheet

For more information, please visit our website at www.cdc.gov/blooddisorders.



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