

DIAMOND BLACKFAN ANAEMIA (DBA)

Key facts

- Diamond Blackfan Anaemia is a rare bone marrow disorder (Approx. only 125 cases in the UK).
- Usually diagnosed before 12 months, but sometimes it can be up to 2 years.
- DBA patients fail to produce red blood cells properly.
- Research has shown a genetic link between ribosomal proteins and DBA, though only around 50% of DBA cases have a known genetic cause.
- In some cases the genetic “error” occurs sporadically rather than being inherited.
- DBA can cause fatigue, poor growth, lack of appetite and a pale complexion.
- Some affected children are short for their age and may have delayed puberty.
- DBA patients also have a slightly elevated risk of certain types of cancer.
- Around half of patients with DBA also have some congenital abnormality. The most common anomalies involve the head and face, arms and hands, heart, and genitourinary tract (including the kidneys, urinary tract, and genital organs)

Aspects of development that can be affected:

- Poor growth
- Delayed puberty
- Elevated risk of cancer
- Increased chance of congenital abnormality

School implications to consider

Strengths

- Condition should not directly affect academic achievement

Considerable weaknesses

- Attendance can be poor as children attend regular transfusions/appointments
- Attention may be affected by fatigue.

Treatment

- Treatment ranges from monthly blood transfusions to regular steroid treatment, and in some cases bone marrow transplant.
- Frequent transfusions cause the accumulation of iron in the body so they must take a ‘chelation’ drug to remove excess iron from their bodies. Often this takes the form of a nightly infusion of medication (Desferal) via a subcutaneous needle over a ten hour period. In other cases an oral drug (Exjade) can be taken daily.
- Steroid treatment can stimulate a response in the bone marrow and instigate red cell production. In these cases transfusions can be reduced in frequency or stopped altogether.
- Bone Marrow Transplant.

Useful websites

<http://diamondblackfan.org.uk/>

The UK based Diamond Blackfan Anaemia Charity. The website contains lots of information and links.

<https://dbafoundation.org/>

The American based Diamond Blackfan Foundation Inc. the website has additional information and links.