

Checklist

Annual check-up for transfused patients with Diamond-Blackfan anaemia (DBA)

Note: Some of the tests below should be repeated at much closer intervals for transfused patients, please discuss this with your attending doctor.

- Patient history: transfusion reactions? side effects of iron chelation therapy ? (auditory/visual disorders, gastro-intestinal issues, skin reactions with subcutaneous iron chelation therapy)
- Full physical examination
- Review of therapeutic goal: Hb levels prior to transfusion > Hb target level (recommended 9.5-10 g/dL)
- Review of annual transfusion volume (target level \leq 250 mL/kg body weight/year)
- Review of iron chelation therapy dose: ferritin value increasing? side effects?
- Lab work:
 - differential blood count including reticulocyte count
 - all electrolytes, urea:creatinine ratio, GPT/gGT/AP/bilirubin/cholinest/protein, ferritin, transferrin saturation, lipase, coagulation
 - immunoglobulin G, *in case of susceptibility to infections/major infections only:* lymphocyte phenotyping
 - antibody screening test (transfusion medicine)
 - urine diagnostics using urine drip test and determination of Na, Ca, Ph, protein, albumin, creatine, protein:creatinine ratio, tubular reabsorption of phosphate
- Serological testing for hepatitis B (HBsAg and anti-HBs quantitative), hepatitis C (anti-HCV), HIV (HIV combotest p24/IgG)
- Endocrinology:
 - parathyroid hormone + Vitamin D
 - TSH/FT3/FT4
 - basal cortisol
 - *from 10th year of life:* fasting blood glucose + insulin (HOMA-IR score)
 - *from 10th year of life:* 24-hour cortisol urine test
 - *from 10th year of life:* oral glucose tolerance test, LH/FSH + testosterone/estradiol, prolactin
 - *in restricted growth/below-percentile growth:* IGF-1/IGFBP-3
- ECG, echocardiography (cardiac insufficiency? pulmonary hypertension?) every two years until 10th year of life, then annually
 - *from 16th year of life:* where necessary, long-term ECG, cardiac MRI (for severe iron overload)
- abdominal ultrasound (particularly urolithiasis/cholelithiasis, splenomegaly)
- annual bone age analysis
- *from 16th year of life:* bone density assessment (DXA) every two years
- eye examination (fundus, visual acuity, colour vision, slit lamp)
- audiometry (hearing test)
- organ iron assessment by MRI of liver/spleen/heart (T2*-weighted imaging)