Pregnancy in Diamond Blackfan Anaemia



The following information has been provided by -Dr. Sarah Ball, Reader/Hon Consultant in Paediatric Haematology St George's Hospital Medical School, London.

This information is not intended to be a substitute for advice from a patients own consultant, as each case of DBA has unique variations. Rather it should be used to provide a starting point for these discussions.

It has been known for some time that pregnancy may be difficult for women with DBA, with respect to the treatment of their own anaemia, but also because of a higher risk of miscarriage. Because it is very unlikely that an obstetrician or midwife will have ever looked after a mother with DBA before, it can be hard for them to know what to do. There has been an important paper just published, summarised below, which makes an excellent starting point for a woman with DBA to discuss the management of pregnancy with her doctors. It is strongly recommended to start these discussions **before** trying for a baby.

Faivre et al 2006 Haematological 91:530-533

High-risk pregnancies in Diamond-Blackfan anemia: a survey of 64 pregnancies from the French and German registries

This reports a joint study from the French and German DBA Registries, looking at 64 pregnancies in women with DBA. The study included 60 women in total, of whom 26 became pregnant. There was a high risk of complications in both mothers and babies. Their important findings were:

- Women with treatment-free DBA (off steroids) were more likely to achieve pregnancy, but still with a risk of complications.
- The 64 pregnancies resulted in only 34 liveborn babies (56%)
- Only 22 of the total 64 pregnancies were uncomplicated (defined as normal maternal blood pressure throughout, delivery after 36 weeks, birth weight above 10th centile, no fetal malformations, but not counting DBA in the baby)
- Prominent complications were pre-eclampsia, intrauterine growth retardation and in utero fetal death
- Complicated and uncomplicated pregnancies could happen sequentially in the same individual
- 13 of the 34 liveborn babies were DBA affected (41%), which fits with the expected risk of 50%
- There was one case of hydrops fetalis, caused by very severe anaemia in a DBA affected fetus
- There did not seem to be an increased risk of complications in pregnancies where the baby was DBA affected
- 8 of the babies of DBA mothers had malformations; this was more likely if the baby had DBA, but a risk even if the baby did not have DBA
- 5 of the 15 women who were treatment independent at the start of pregnancy needed transfusion or steroids during their pregnancy
- 6 of the 11 women already on treatment needed extra steroid or more frequent transfusions to control anaemia during pregnancy (no woman in this study showed the prolactin effect, in which anaemia improves in late pregnancy)

Their recommendations were:

- It is mandatory that pregnancy should be managed in an obstetric unit with close working between the obstetrician and a haematologist with experience in DBA
- The pregnancy should be closely monitored with regular ultrasound scans, including Doppler, to monitor placental and fetal development
- Aim to keep Hb above 10g/dl during pregnancy (this could mean an increase in steroids, but avoid pushing the dose too high; better to have the occasional transfusion)
- Consider periconceptual low dose aspirin (ie start while trying for a baby, like folic acid), continued up to 37 weeks gestation, especially in women with a previous miscarriage.

There are other factors to consider (not covered in the above paper):

- Iron chelation therapy (whether desferrioxamine, deferiprone or Exjade) may harm unborn babies, which is very important for women with DBA who are on transfusions and chelation. The safest approach is to make sure that iron levels are really well controlled before trying for a baby, to allow an interruption in chelation
- There is very little reported about pregnancies in which the father rather than the mother has DBA, except that there will be a 50% risk that the baby will be affected by DBA