

Newcastle Mitochondrial Centre

At a glance guidelines:

Pregnancy in Mitochondrial Disease

For full guideline visit:

<http://www.newcastle-mitochondria.com/service/patient-care-guidelines/>

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There are many different forms of mitochondrial disease, varying greatly in their genetic basis, clinical presentation, progression and prognosis. We recommend referral to a specialist mitochondrial centre for appropriate counselling and guidance (www.mitochondrialncg.nhs.uk or see appendix for international centres).

Pregnancy in patients with mitochondrial disease is usually well tolerated, however there does appear to be an increased risk of complications such as gestational diabetes, pre-eclampsia, and preterm delivery. This is predominantly seen in patients carrying the m.3243A>G mtDNA mutation.

We therefore recommend the following:

1. **Pre-conception counselling:** all women at risk from mitochondrial disease who are considering pregnancy should be seen in a specialist mitochondrial disease clinic for pre-conception counselling (irrespective of genotype or symptoms).
2. **Medication review:** this should include a discussion regarding potential risks and benefits of current medications (eg. AEDs, ACE inhibitors) in the context of mitochondrial disease.
3. **Folic acid:** supplements should be used in accordance with the NICE guidelines (CG62) and prescribed at 5mg/day for women with diabetes (CG63) or those taking anti-epileptic drugs (CG137).

4. **Obstetric assessment:** all women with mitochondrial disease who become pregnant should have an early initial obstetric assessment and close monitoring throughout their pregnancy.

5. **Gestational diabetes (GDM):** mitochondrial disease increases the risks of gestational diabetes. We recommend an oral glucose tolerance test (OGTT) as follows:
 - 6.1. at 16 weeks gestation for women with a previous history of GDM and again at 24-28 weeks if the 16 week OGTT was normal (www.nice.org.uk/guidelines CG63).
 - 6.2. at 20 weeks gestation for women carrying mutations with a high risk of diabetes (e.g. m.3243A>G, 14709T>C).
 - 6.3. At 24-28 weeks gestation for women with other risk factors (e.g. BMI > 30kg/m², high prevalence ethnic origin etc) as per NICE guidelines (www.nice.org.uk/guidelines CG63).

6. **Multi-disciplinary care:** all women should have access to multi-disciplinary care between obstetrics, mitochondrial medicine and other medical specialties as indicated. Cardiac or respiratory compromise may develop in patients with advanced multisystem involvement and liaison with appropriate specialists is advised from the outset.

7. **Aspirin and pre-eclampsia:** women with mitochondrial disease should only receive aspirin (from 12 weeks until

delivery) if they have previously suffered pre-eclampsia. This is the same as other women considered to be at high risk of recurrence (NICE Hypertension in pregnancy guideline CG 107).

8. **Magnesium sulphate in pre-eclampsia:** if pre-eclampsia develops magnesium sulphate infusion may be used to prevent progression to eclampsia but vigilance for magnesium toxicity is advised.