An update from Dr Woods on pre-natal screening

A few families have asked if prenatal diagnosis is possible, if they already have a child with Microcephaly. Dr Woods emailed us this reply.

For prenatal testing a family has to go through a clinical genetics department. There needs to be a diagnosis made as to the cause of microcephaly. From this the Clinical Geneticist will know the chance (or range of chances) of the condition recurring in future pregnancies. Also they will know if prenatal testing is possible. And, if it is,how? This may be by ultrasound scan, chromosome analysis or sometimes DNA testing for a specific gene defect. Thinking of autosomal recessive primary microcephaly, there are at least 8 genes, but thank heavens, about 1/2 of cases are caused by faults (mutations) in the MCPH5 gene ASPM. ASPM testing is done by my lab on a research basis - but soon will be offered by the Cambridge NHS DNA lab as a Gene Dossier test (that is, agreed by government that the test should be paid for by the PCT/funders). We only accept samples for analysis from clinical geneticists and reject quite a few as they don't meet our diagnostic criteria.

The main thing here is to specifically see a clinical geneticist about the child's CAUSE of microcephaly. If this can be determined, then prenatal may be possible.

Best wishes from Dr Woods