Neural Tube Defects (NTDs) and genetic testing
Information for PARENTS

Do all patients with a neural tube defect (NTD) need to be referred to Clinical Genetics?

No, most patients with an isolated neural tube defect (i.e. spina bifida or anencephaly occurring without other features) do **not** need to be seen by Clinical Genetics.

However, if there are additional features (e.g. other congenital anomalies, or a strong family history of neural tube defects), these patients may benefit from a referral to Clinical Genetics.

Is genetic testing recommended for neural tube defects?

Genetic testing is **not** routinely recommended for isolated neural tube defects. The genetics of isolated neural tube defects remains poorly understood, and at present, we believe they may be associated with complex genetic (polygenic) and environmental factors.

Genetic testing may be indicated in the case of neural tube defects occurring as part of a syndrome (i.e. a collection of symptoms which occur together as part of an underlying genetic diagnosis).

What is the risk of recurrence in a future pregnancy, and what supplements are recommended?

Individuals who have had one pregnancy affected by a neural tube defect have an increased risk of this happening again (estimated ~3% recurrence risk). The recurrence risk may be higher, if you have had more than one child with a neural tube defect, or if the neural tube defect has occurred as part of a syndrome.

You can reduce the risk of recurrence by taking high dose folic acid (5mg), prescribed by your GP, in future pregnancies (at least one month before conception, and for the first 12 weeks of pregnancy). This advice would also apply to any of your close female relations (e.g. your sisters, or the female partners of your brothers).

Is *MTHFR* gene testing recommended?

No. Testing of the *MTHFR* gene is not currently recommended in the case of isolated neural tube defects.

Some private companies offer direct-to-consumer testing of *MTHFR*. There are some variants or 'polymorphisms' in *MTHFR,* which are commonly found in the general population. It is estimated that ~10% of individuals in the UK & Ireland are homozygous for the C667T variant ("TT homozygous"). This means they have the C667T variant on both copies of their *MTHFR* gene. A1298C is another commonly observed *MTHFR* 'polymorphism'. At present, there are no specific recommendations for women who are homozygous carriers for these common variants, and we would reassure these individuals that, based on the scientific evidence currently available, we would not expect them to be at a significantly increased risk of having a child with a neural tube defect, and no specific supplements are recommended.

Other useful resources:

https://www.shinecharity.org.uk/
https://www.gov.uk/government/publications/anencephaly-description-in-brief/anencephaly-information-for-parents
https://www.gov.uk/government/publications/spina-bifida-information-for-parents