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Important points

- Some genetic conditions are caused by an interaction between the inherited genetic information and the environment such as diet and lifestyle factors. If exposure to the environmental trigger can be avoided, it may be possible to prevent the person being affected with the condition despite their genetic make-up
- In the majority of cases of neural tube defects, a multifactorial condition that affects the development of the spine and brain in babies, the genetic contribution is still to be defined but the environmental trigger has been identified as a lack of the vitamin folic acid (or folate) in the mother i.e. in the uterine environment in which the fetus is developing
- It is estimated that seven out of ten (70%) cases of neural tube defects can be prevented by increasing the mother's intake of the B group vitamin called folate or folic acid prior to, and during early pregnancy. Folate is naturally found in leafy green vegetables, wholegrain breads, cereals, legumes such as peas, dried beans and lentils. It is also available in tablet form as folic acid
- It is recommended that for all women folate levels should be
 - High for at least one month prior to possible conception **and**
 - Continued at that level for the first three months of pregnancy
- **For most women** it is necessary to have about 0.4mg to 0.5mg (milligrams) of folate each day
- **Women who are at increased risk** will need to have more than 0.5mg each day as advised by their doctor
- Women are at increased risk if:
 - They have had a baby with spina bifida, anencephaly or other neural tube defect(s)
 - They themselves were born with a neural tube defect
 - They have a close relative born with a neural tube defect
- Women taking drugs to control epilepsy or seizures should take folic acid only as advised by their doctor
- There are tests available during pregnancy to check for spina bifida or other neural tube defects in a baby

Some genetic conditions are caused by an interaction between the inherited genetic information and the environment such as diet and lifestyle factors.

Such conditions are referred to as '**multifactorial**' conditions (see Genetics Fact Sheet 11).

An individual will be affected by these conditions if their genetic information means that they are predisposed to the condition, and they are exposed to the environmental factor that can 'trigger' it.

If exposure to the environmental trigger can be prevented, it may be possible to prevent the person being affected with the condition despite their genetic make-up.

Can all multifactorial conditions be prevented?

No. The environmental trigger is not known for most of these conditions.

In the case of neural tube defects, however, the environmental trigger has been identified as a lack of folate in the mother in most cases (see Genetics Fact Sheet 59).

Prevention of a genetic condition - neural tube defects

Spina bifida and anencephaly are problems with the development of the spine or brain, and are also classified as neural tube defects. They

- Occur very early in pregnancy, by the 28th day after conception. This is often before a woman knows she is pregnant

- Cause many babies to die and others to have problems with walking and with bowel and bladder control
- Affect about 1 in every 500 pregnancies
- Affect about 1 in every 600 births – the difference is due to the loss of pregnancies affected with neural tube defects

In the majority of cases of neural tube defects the genetic contribution is still to be defined but the environmental trigger in this condition has been identified as a lack of the vitamin folic acid in the mother i.e. in the uterine environment in which the fetus is developing.

- Having enough of a simple vitamin called folic acid or folate can help prevent babies being affected with neural tube defects
- Research has shown that 7 out of 10 (70%) cases of neural tube defects can be prevented by increasing the woman's intake of the vitamin folate or folic acid prior to, and during early pregnancy

It is recommended that for all women folate levels should be high for a least one month prior to possible conception **and** continued at that level for the first three months of pregnancy.

This timing is advised since many people are not able to accurately determine the time of conception.

A woman often does not know she is pregnant in the first four weeks after conception, by which time the neural tube will have closed. Therefore, after this time taking folate is unlikely to prevent a neural tube defect in a baby.

What is folate?

Folate is a B group vitamin naturally found in

- Leafy green vegetables
- Wholegrain breads
- Cereals
- Legumes such as peas, dried beans and lentils

Some foods are supplemented with folate. The addition of folate to certain foods in Australia has been permitted since June 1995

- A number of cereals, breads and juices are supplemented with folate
- Check labels to identify food sources of folate

Folate is also available in tablet form as folic acid

Who needs folate?

All women - especially those women planning a pregnancy.

As many pregnancies are unplanned, all women of reproductive age should make sure that they have a folate rich diet or take a folic acid tablet each day.

Folate, however, will only help in preventing spina bifida or other neural tube defects in babies if it is being taken around the time of conception and for the first twenty-eight days of pregnancy, or until the neural tube has completely formed.

A folate rich diet based on a wide variety of vegetables, fruit, legumes, wholegrain breads and cereals is a healthy way of eating for everyone. This way of eating can help prevent heart disease, some cancers, diabetes and other diseases.

How much folate is needed before, and early in pregnancy?

For most women

- It is necessary to have about 0.4mg to 0.5mg (milligrams) of folate each day
- This can be obtained by:
 - Eating a folate rich diet (consult your doctor, nutritionist or genetic counsellor)
 - Taking a daily low dose folic acid tablet (0.5mg) available from chemists, health food stores and supermarkets

Should all women have 0.5 mg folate each day?

In general, yes.

Some women, however, are at higher risk than the average woman of having a baby with a neural tube defect.

These women will require more than 0.5mg of folate daily before and during early pregnancy.

Women are at increased risk if:

- They have had a baby with spina bifida, anencephaly or other neural tube defect
- They themselves were born with a neural tube defect
- They have a close relative who was born with a neural tube defect

These women should consult their doctor or genetic counsellor before pregnancy for advice regarding the amount of folate they should take.

A note of caution about taking folate:

Women taking drugs to control epilepsy or seizures should take folic acid only as advised by their doctor.

Multi-vitamin and mineral tablets should not be taken in high doses to get the right amount of folic acid as one of the other vitamins could be too high and cause other problems with the baby. This is particularly so for vitamin A that has been shown to cause serious birth defects in babies when taken in high doses.

There is some evidence that multi-vitamin tablets taken in low doses may reduce the risk of other birth defects. It is important to discuss the use of such tablets with your physician or obstetrician **before** becoming pregnant.

Other benefits of folate

There is some evidence that folate can also reduce the incidence of cleft lip with or without cleft palate. This applies to situations in which the cleft lip and/or palate occurs alone, without any other physical symptoms.

Testing in pregnancy for spina bifida and other neural tube defects

There are tests available during pregnancy such as specialised ultrasound tests to check for spina bifida or other neural tube defects in a baby.

See Genetics Fact Sheet 17 for a full discussion about prenatal testing.

Other Genetics Fact Sheets referred to in this Fact Sheet: 11, 17, 59

Information in this Fact Sheet is sourced from:

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