

Family Planning For Couples Affected By 5q SMA

You can also read this guide on our website at smauk.org.uk/8nvk where you can follow all the links we give to further information.

5q Spinal Muscular Atrophy is the most common form of SMA. It includes SMA Types 1, 2, 3 and 4.

Who this is for

We hope this information will be helpful for couples::

- who have had a child with 5q SMA
- who have not had a child with 5q SMA but know that one or both are carriers of 5q SMA
- where one or both have 5q SMA
- where someone in one or both of their extended families has 5q SMA, but they do not know if they carry the condition.

Please be aware that your options and any funding may be limited by your genetic, family and individual circumstances.

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1. Inheriting 5q SMA

The chances of having a baby with 5q SMA vary depending on whether you or your partner have 5q SMA or, are carriers.

5q SMA is confirmed via a test for the *SMN1* gene. The test cannot predict the 'Type' of SMA. To find out how severe the symptoms might be, the genetics laboratory must also test the *SMN2* copy numbers.

If you already have a child who has 5q SMA and your unborn baby inherits it too, they are likely to have the same Type of 5q SMA. 5q SMA tends to affect siblings in a similar way, or at a similar age. But this is not always the case.

For more information see: [The Genetics of 5q SMA >](#).

2. Genetic Counselling

A Genetic Counsellor can discuss your individual genetic circumstances with you. They can give advice and information about all your testing options. This can be before you consider any pregnancy or as early in your pregnancy as possible.

If it's possible, Genetic Counselling before pregnancy can help you and your partner consider genetic testing. It can give you time to think about the difficult decisions this can raise. Genetic Counsellors will support you through the emotional impact of any option you choose. You will be able to discuss what the next steps might be for you and your family.

If you have a child who has SMA: you should have already been referred for Genetic Counselling. If you are thinking about a further pregnancy, you should be able to return to them for further advice and support.

If you have SMA or are a carrier of SMA: you can be referred to your local Genetics Centre by a GP, specialist, midwife or obstetrician. If you are pregnant, you can ask for an urgent Genetic Counselling referral. If this isn't possible, you can contact your local genetic service directly.

If someone in your extended family has SMA, but you do not know if you are a carrier: talk to your GP. Ask them to refer you and your partner to your regional Genetics Centre for genetic testing.

In general, genetic testing cannot guarantee 100% accuracy. However, steps are taken to make sure results are as close to 100% accuracy as possible.

For more information see [The Genetics of 5q SMA >](#).

To find your nearest genetic service see:

- [Genetic Alliance UK >](#)
- [British Society for Genetic Medicine >](#)

For more general information about genetic services and testing:

- [Genetic Alliance UK >](#)

3. Options for Testing Before or During Pregnancy

You may wish to consider testing a pregnancy for SMA if:

- you and your partner are both carriers of SMA *or*
- one of you has SMA and the other is a carrier.

There are various reasons couples test for SMA. They might want to prepare for their baby's arrival. Or they may decide to end the pregnancy if the pregnancy is predicted to be affected by SMA.

There is a chance that a child will inherit 5q SMA if:

- both partners are carriers of SMA **or**
- one partner has SMA; the other is a carrier.

For more information see [The Genetics of 5q SMA >](#).

The following family planning options may be available to you:

- Conceive naturally and avoid any testing for SMA during a pregnancy. Your Genetic Counsellor can discuss the chances of you having a child who has SMA. If you wish, they can organise testing for the baby when they are born.
- Have a type of fertility treatment which tests the embryos for SMA **before** achieving a pregnancy.
- Conceive naturally and have testing **during** pregnancy to see if the baby will have SMA.

These options are outlined on the next pages.

4. Conceiving naturally and testing at birth

Newborn screening for SMA is not yet routine in the UK. Genetic testing for SMA at birth is not usually offered if:

- a baby shows no symptoms **and**
- only one parent is a carrier of SMA **or**
- neither parent is a carrier of SMA.

If both parents are carriers of SMA, you can ask your Genetic Counsellor if your newborn baby can be tested. This is usually done by taking a cord blood sample, or a blood sample directly from the baby. Both can be done shortly after the birth of the baby. The results are usually available within 1-2 weeks. It sometimes takes longer.

If the test shows SMA is suspected, your baby will be seen urgently by a specialist neuromuscular consultant. They will discuss drug treatment options. The treatments work best if started before there is any muscle weakness, or when this is minimal. It is therefore important for treatment to be started as soon as possible.

See:

[**Drug Treatments for Children who have 5q SMA – England, Wales & Northern Ireland >**](#)

[**Drug Treatments for Children who have 5q SMA – Scotland >**](#)

These tell you about the treatments, how they work and which children may have access.

It may be helpful to talk about what the treatment options would be. You can do this with your Genetic Counsellor, or clinical team if you have one. Depending on your circumstances, this could be before you conceive naturally or early on in your pregnancy. Your Genetic Counsellor can contact the nearest [**Regional Neuromuscular Specialist Centre >**](#) for latest information.

5. In Vitro Fertilisation (IVF) – with Preimplantation Genetic Testing for Monogenic/Single Gene Defects (PGT-M)

- **Who can have IVF with PGT-M?**

In the United Kingdom, the Human Fertilisation and Embryology Authority (HFEA) has approved the use of PGT-M for couples at risk of having a child with SMA Types 1, 2 or 3.

This may be an option **before** pregnancy. It is primarily for couples who want to exclude the possibility of their baby having 5q SMA

There are eligibility criteria for both the PGT-M process and for NHS funding. These overlap. Different PGT-M centres may have different criteria. Your Genetic Counsellor will explain whether you are eligible.

- **What does IVF with PGT-M involve?**

In-vitro fertilisation (IVF) is carried out using a couple's own eggs and sperm. Eggs are removed from the woman's ovaries and fertilised with sperm in a laboratory.

After about 5 days, the embryos grow into balls of cells called "blastocysts." A small sample of cells is carefully taken from each one. This is known as an embryo "biopsy." After the biopsy, the embryos are quickly frozen using a method called 'vitrification'. This happens while the cells are tested for 5q SMA.

If the genetic test results show at least one embryo without SMA, the medical team can select, warm, and transfer that embryo to the womb.

A pregnancy test is carried out 12 days later to see whether the PGT-M treatment has been successful.

- **How long does it take to get started?**

There is often a wait for the initial consultation. After that, starting PGT-M may be delayed while ethical approval and / or funding is arranged.

PGT-M also involves IVF (In vitro fertilisation). This also takes time. Realistically PGT-M may take many months, or sometimes even years.

To discuss this option with a Genetic Counsellor, ask for a referral to your local Regional Genetics clinic. To find your nearest genetic service see:

- [Genetic Alliance UK >](#)
- [British Society for Genetic Medicine >](#)

For more information about PGT-M, see: [Genetic Alliance UK >](#)

6. Non-Invasive Prenatal Diagnosis (NIPD)

• Who can have NIPD?

This is an option for couples who wish to know whether their baby has inherited SMA. It usually takes place **from around the 9th week of pregnancy**. This option does not involve invasive testing. There is no risk of miscarriage because of the test.

To have the test:

- Both parents must be known carriers of an *SMN1* gene alteration **and**
- A DNA sample must be available from:
 - the family member who has SMA. This may be a previous child of the couple, or another relative who has SMA. If a couple has a child who has SMA, there are probably lab samples from earlier tests. A new sample may not be needed **or**
 - a previous child of the couple who has been confirmed not to be a carrier. They won't have an altered copy of the *SMN1* gene.

Testing can be done in single pregnancies. It may not be possible if there is more than one fetus. This test is not currently possible for couples who are related (consanguineous).

• What does NIPD involve?

A sample of the mother's blood is taken from when they are around 9 weeks pregnant. This is tested for the unborn baby's genetic material.

The test result should be available within 21 days. Results are often available more quickly than this.

• What will the NIPD Test Show?

The NIPD testing will show whether the fetus has inherited the affected or unaffected copies of the *SMN1* gene from each parent.

- If the fetus has inherited one affected copy of the *SMN1* gene, and one unaffected copy, they are a carrier of 5q SMA. They are very unlikely to have SMA.
- If the fetus has inherited two affected copies of the *SMN1* gene – one from each parent – the child will have 5q SMA.

This testing will not check for *SMN2* copy numbers which help predict how severe the SMA symptoms might be.

If you already have a child who has 5q SMA and your unborn baby inherits it too, they are likely to have the same Type of 5q SMA. 5q SMA tends to affect siblings in a similar way, or at a similar age. But this is not always the case.

For more information see: [The Genetics of 5q SMA](#) >.

Very occasionally, the test may not return a result. If this happens the test may be repeated or an alternative test may be carried out. See CVS and amniocentesis below.

When using NIPD, there is a small, estimated risk of misdiagnosis. This is approximately 1 in every 500-1000 diagnoses. This is due to technical and biological reasons.

• What Next?

You would be given information and support before, during and after your NIPD treatment. This will help you decide what to do next. Your options would be to continue with the pregnancy or, depending on the stage of your pregnancy, to end the pregnancy.

If you have previously had a child diagnosed with SMA Type 1 you may wish to listen to our webinar on [Treatment during Pregnancy for a baby diagnosed with SMA Type 1 >](#). You can then ask your child's neuromuscular consultant if this could be an option if your NIPD test shows the fetus has SMA. Please be aware this treatment is not yet part of standard NHS care.

For information about ending a pregnancy, see:

- [The NHS Website >](#)
- [Antenatal Results and Choices >](#)

• How can I access this test?

Access to this testing and Genetic Counselling is through your local genetics centre. NHS funding is available for this test when both parents are confirmed carriers of SMA. Your genetics centre will be able to advise you on eligibility for NIPD.

To find your nearest genetic service see:

- [Genetic Alliance UK >](#)
- [British Society for Genetic Medicine >](#)

7. Chorionic Villus Sampling (CVS)

- **Who can have CVS?**

This is an option for couples who wish to know if their baby has SMA before birth. It is usually carried out **between the 11th and 14th weeks of pregnancy**. If necessary, it is sometimes performed later than this.

- **What does CVS involve?**

CVS involves taking a sample of the placenta and testing the cells for 5q SMA. There are two options: inserting a needle through the abdomen or inserting a tube through the cervix. Both processes are guided by an ultrasound scan.

The sample is sent to a laboratory and genetically tested for 5q SMA. It generally takes a week for the test result, but this may vary. You can ask how long it will take for the result and decide how you will be informed.

The laboratory may also test for SMN2 copy numbers. This can help your genetics team to predict how severe the SMA symptoms might be.

If you already have a child who has 5q SMA and your unborn baby inherits it too, they are likely to have the same Type of 5q SMA. 5q SMA tends to affect siblings in a similar way, or at a similar age. But this is not always the case.

- For more information see: [The Genetics of 5q SMA >](#)

Rarely, a CVS can cause miscarriage, the loss of the pregnancy in the first 23 weeks. The chance of miscarrying after CVS is thought to be less than 1 in 200 for most pregnancies, and around 1 in 100 for multiple pregnancies (such as twins).

- For more information about CVS, see the [NHS website >](#)

• What Next?

You would be given information and support before, during and after your CVS treatment to help you decide what to do next. Your options at this point would be to continue with the pregnancy or end the pregnancy. There are different ways to do this depending on the stage of the pregnancy.

If you have previously had a child diagnosed with SMA Type 1 you may wish to listen to our webinar on [Treatment during Pregnancy for a baby diagnosed with SMA Type 1](#) >. You can then ask your child's neuromuscular consultant if this could be an option if your NIPD test shows the fetus has SMA. Please be aware this treatment is not yet part of standard NHS care.

For information about ending a pregnancy, see:

- [The NHS Website](#) >
- [Antenatal Results and Choices](#) >

8. Amniocentesis

This is an option for couples who wish to know whether their baby has SMA during a pregnancy. It usually takes place **between the 15th and 20th weeks of pregnancy**

- **What does amniocentesis involve?**

A needle is put through the abdomen to remove some of the fluid that surrounds the fetus in the womb – the amniotic fluid. The process is guided by an ultrasound scan. The cells in the amniotic fluid are sent to a laboratory and genetically tested for 5q SMA.

The laboratory may also test for SMN2 copy numbers. This can help your genetics team to predict how severe the SMA symptoms might be.

If you already have a child who has 5q SMA and your unborn baby inherits it too, they are likely to have the same Type of 5q SMA. 5q SMA tends to affect siblings in a similar way, or at a similar age. But this is not always the case.

Rarely, amniocentesis can cause miscarriage – the loss of the pregnancy in the first 23 weeks. This is estimated to occur in up to 1 out of every 200 women who have amniocentesis. The risk of amniocentesis causing complications is higher if it is carried out before the 15th week of pregnancy. This is why the test is only done after this point.

- **For more information see: [the NHS website](#) ›**

- **What Next?**

You would be given information and support before, during and after your amniocentesis to help you decide what to do next. Your options at this point would be to continue with the pregnancy or to end the pregnancy. There are different ways to do this depending on the stage of the pregnancy.

If you have previously had a child diagnosed with SMA Type 1 you may wish to listen to our webinar on [Treatment during Pregnancy for a baby diagnosed with SMA Type 1 >](#). You can then ask your child's neuromuscular consultant if this could be an option if your NIPD test shows the fetus has SMA. Please be aware this treatment is not yet part of standard NHS care.

For more information about ending a pregnancy, see:

- [The NHS Website >](#)
- [Antenatal Results and Choices >](#)

9. In Vitro Fertilisation (IVF) – with Egg or Sperm Donation

This is an option **before pregnancy**. It may be available for couples who:

- want to exclude the possibility of their baby having 5q SMA **and / or**
- may be having difficulty conceiving.

• What does IVF involve?

During IVF, an egg is removed from a woman's ovaries and fertilised with sperm in a laboratory. After a few days, the fertilised egg, called an embryo, is then transferred into the prospective mother's womb. There is then a 2 week wait for a pregnancy test to see if the treatment has worked.

The IVF would use eggs and / or sperm from donors who have been tested to check they are not carriers of 5q SMA.

Your Genetic Counsellor will be able to discuss whether this could be an option for you.

To find your nearest genetic service see:

- [Genetic Alliance UK >](#)
- [British Society for Genetic Medicine >](#)

For more information about IVF, see [the NHS website >](#). But please note the funding and eligibility set out in these pages is for couples who have fertility challenges.

10. Sources Of Support

• Antenatal Results and Choices (ARC)

ARC > ARC is a national charity. It helps parents and healthcare professionals with antenatal screening and its effects. They offer non-directive information and support to parents:

- before, during and after antenatal screening
- if their baby has a diagnosis
- if they are making difficult decisions about continuing with or ending a pregnancy
- if they are coping with complex and painful issues after making a decision, including bereavement.

Helpline: 0207 713 7486

Email: info@arc-uk.org

Text: 07908 683004

• Miscarriage Association

The **Miscarriage Association** > information and support for anyone affected by miscarriage. Helpline: 01924 200 799

• SMA UK

We recommend you talk with a Genetic Counsellor. If after this you would like further support, you can talk with our support team. We are not experts in family planning options but do provide free, confidential information and support to anyone affected by SMA. **Phone: 01789 267520** or [contact our Support Team](#) >.

Or you can make a request for confidential emotional counselling with our partner service [Rareminds](#) >.



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www.smauk.org.uk • office@smauk.org.uk • 01789 267520

The Mansley Business Centre, Timothy's Bridge Road, Stratford-upon-Avon, CV37 9NQ

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