5q Spinal Muscular Atrophy (5q SMA) is the most common form of SMA; it includes SMA Types 1, 2, 3 and 4. We hope this guide will be helpful for couples who are thinking of having children or who are currently pregnant:

➢ who have had a child with 5q SMA
➢ who know that one or both of them are carriers of 5q SMA
➢ where one or both of them have 5q SMA
➢ where someone in one or both of their extended families has 5q SMA, but they don’t know if they are carriers of the condition.

Please be aware as you read this that the different options available to you, and funding for them, may be limited by your particular genetic, family and individual circumstances.

The decision about which option to choose is very personal; there are no right or wrong choices. You’ll want to make your own decision, based on the information given to you by the health professionals supporting you.
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.

For more information about this you may want to read:

➢ The Genetics of 5q SMA
www.smauk.org.uk/the-genetics-of-5q-sma

If you already have a child with 5q SMA and your unborn baby also has 5q SMA then it’s likely, but not certain, that your unborn baby will also have the same Type of 5q SMA.

Genetic Counselling

A genetic counsellor can discuss your individual genetic circumstances with you and give advice and information about all your testing options either before you consider any pregnancy or as early in your pregnancy as possible. If possible, genetic counselling before pregnancy will give you and your partner more time to think about genetic testing and the possibly difficult decisions this can raise. Genetic counselling will also support you through the emotional impact of any option you choose and discuss what the next steps might be for you and your family.

If SMA has occurred in your extended family but you don’t know if you are a carrier of SMA and are thinking of having children, you can ask your GP to refer you and your partner to your regional Genetics Centre for genetic testing.

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

Some couples choose not to have any tests either before or during a pregnancy.

If you choose this option, you may still want to consider requesting that your baby is genetically tested for 5q SMA shortly after birth. You can ask your medical team or genetic counsellor for more information about this possibility.

In vitro fertilisation (IVF) - egg or sperm donation

This is an option before pregnancy which 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 it 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.

Depending on a couple’s circumstances, IVF would be carried out using eggs and / or sperm from donors who have been tested to check that they are not carriers of 5q SMA.

Your genetic counsellor will be able to discuss whether this could be an option for you.

For more information about what IVF involves, see NHS website:

www.nhs.uk/conditions/ivf

but please note the funding and eligibility set out in these pages is for couples who have fertility challenges.
Preimplantation Genetic Diagnosis (PGD)

This may be an option before pregnancy which may be available for couples who want to exclude the possibility of their baby having 5q SMA and / or may be having difficulty conceiving. There is often a wait for the initial consultation and then starting PGD may be delayed while ethical approval and / or funding is arranged. As PGD also involves IVF (In vitro fertilisation) and this also takes time, realistically PGD may take many months, or sometimes even years.

What does it involve?

Couples use their own eggs and sperm but via IVF, even if they can become pregnant naturally. Eggs are removed from the woman’s ovaries and fertilised with a sperm in a laboratory. This is so that embryos can be tested for 5q SMA. This takes around two weeks to complete.

Once the embryos grow into balls of cells (‘blastocysts’), a small sample of cells is very carefully removed (an embryo ‘biopsy’). After the biopsy, the embryos are rapidly frozen (vitrification) while cells are genetically tested.

Once the genetic test is completed, and providing the results show that there is at least one embryo that doesn’t have SMA, this embryo can then be selected, warmed and transferred into the womb. A pregnancy test is carried out 12 days later to see whether the PGD treatment has been successful.

Who can have PGD?

There are eligibility criteria for both the PGD process and for NHS funding. These overlap, so some couples may meet one set of criteria but not the other, neither, or both. Different PGD centres may also have different criteria. Your genetic counsellor will explain whether you are eligible.
A personal experience of PGD

“Our son was diagnosed with SMA Type 2 when he was 9 months old. As much as the diagnosis itself was a shock, the thought of potentially having more children with the same condition was devastating for us. After a lot of thought and reading through the various information available we asked to be put on the waiting list for PGD.

The medication routine for the IVF part of the process was relatively simple and much easier than I had thought even though it involved me sticking needles into myself. The part that I was most nervous about beforehand was the egg collection but, with a lot of reassurance from the medical staff at the hospital and sedative medication, it all went very well, and I did not feel any pain at all. I cannot remember details of how many eggs they were able to collect or how many were fertilised but in the end I know there was only one embryo that was healthy and suitable for implantation.

The hardest part of the process however was not the medical procedures that we went through but the emotional rollercoaster ride that I was on during the time I was taking the IVF medication as my hormones and my moods were all over the place.

My husband and I didn’t dare to hope that I would fall pregnant on our first treatment round but that is exactly what happened and finally a little baby girl was born, and we named her Sophia. The journey has not been easy, but our children are worth it.”
Non-Invasive Prenatal Diagnosis (NIPD)

This is an option from around the 8th week of pregnancy for couples who wish to know whether their baby has SMA.

It can be used in single pregnancies where both parents are carriers of the SMN1 gene that causes 5q SMA and have previously had a child with genetically confirmed 5q SMA. It cannot be used for a twin pregnancy.

What’s involved?

Three samples are needed for this test:

➢ The mother’s blood is tested for the unborn baby’s (foetus’) genetic material, known as ‘cell-free fetal DNA’. At this stage of pregnancy, the small amount that is present in her blood is sufficient for the test. This test doesn’t present any risk to the foetus – there is no risk of the test causing a miscarriage.

➢ DNA from the child who already has 5q SMA

➢ DNA from the father

It’s likely that there are already stored laboratory samples from previous testing for both the father and the affected child, in which case new samples from them may not be needed.

The test result could be available within 7-10 working days.

What will the test show?

The NIPD testing will show whether the foetus has inherited the affected or unaffected copy of the SMN1 gene from each parent and whether the foetus has 5q SMA. It cannot show the Type of 5q SMA.

It’s estimated that in approximately 5% of cases, the test may not return a result. If this happens the test may be repeated or an alternative invasive test (CVS or amniocentesis – see next sections) may be carried out if the couple wishes. It’s also estimated that when using NIPD, the risk of misdiagnosis, due to technical and biological reasons, is very low (approximately 1 in 500-1000).
What next?

You would be given information and support before, during and after your NIPD treatment to help you decide what to do next. Your options at this point would be to continue with the pregnancy or, depending on the stage of your pregnancy, to have a termination.

Where can I get the test?

Access to this testing and genetic counselling is through your local genetics centre, wherever you live in the UK. Your genetics centre will be able to advise you on funding / charges for NIPD.

Chorionic Villus Sampling (CVS)

This is an option between the 11th and 14th weeks of pregnancy for couples who wish to know whether their baby has SMA.

What is involved?

CVS involves taking a sample of the placenta and testing the cells for 5q SMA. This involves either inserting a needle through the abdomen (transabdominal CVS) or inserting a tube through the cervix (transcervical CVS). Both processes are guided by an ultrasound scanner.

The sample is sent to a laboratory and genetically tested for 5q SMA. It cannot show the Type of 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.

It’s generally agreed that there is an increased risk of miscarriage associated with CVS, though two sources describe this slightly differently. The NHS website says that the risk of miscarriage after CVS is estimated to be about 0.5 to 1%, though it’s difficult to determine which miscarriages would happen anyway and which are the result of CVS procedure. However, in their guidelines, the Royal College of Obstetricians & Gynaecologists say that women should be informed that the additional risk of miscarriage following CVS may be slightly higher than that of amniocentesis (which is around 1%).

For more information about CVS see:

➢ NHS website: www.nhs.uk/conditions/chorionic-villus-sampling-cvs/
➢ Royal College of Obstetricians & Gynaecologists: www.rcog.org.uk/en/guidelines-research-services/guidelines/gtg8/
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 to have a termination.

For more information about termination of pregnancy, see:
Antenatal Results and Choices:
www.arc-uk.org/for-parents/ending-a-pregnancy

A personal experience of CVS

“I found that the genetics nurse was very helpful in liaising between the laboratory, the hospital and us. It’s worth asking to have a dating scan at around 8 weeks, as this will confirm the dates are approximately correct and reduce the possibility of being disappointed on the arranged date of the CVS test.

My consultant didn’t use a local anaesthetic, as some do, and the first CVS test was quite painful. The second one was merely uncomfortable in comparison - every pregnancy is different.

The CVS results took approximately a week. When we had a bad CVS result it was devastating, but it was still in a way a relief that the waiting was over and we could get on with trying again. I believe that nothing is as bad as losing a child and this enabled me to go through with a termination, knowing that I wouldn’t have to watch another baby die.

In time, we finally had a good CVS result. I was in shock to start with, even though my partner was celebrating as soon as we heard. 6 months later, we got the child we’d been waiting for. Although it will never take away the pain of losing our first born, our new baby has brought joy and happiness back into our lives.”
Amniocentesis

This is an option between the 15th and 20th weeks of pregnancy for couples who wish to know whether their baby has SMA.

What’s involved?

Amniocentesis involves putting a needle through the abdomen to remove some of the fluid that surrounds the foetus in the womb (amniotic fluid).

The cells in the amniotic fluid are sent to a laboratory and genetically tested for 5q SMA. It cannot show the Type of SMA. Amniocentesis test results can take longer than CVS results. It generally takes up to 2 weeks for the test result, but this may vary. You can ask how long it will take for your result and decide how you will be informed.

It’s generally agreed that there is an increased risk of miscarriage associated with amniocentesis, though two sources describe this slightly differently. The NHS website says that if you have amniocentesis after 15 weeks of pregnancy, the chance of having a miscarriage is estimated to be 0.5 – 1%, though the risk is higher if the procedure is carried out before 15 weeks. However, the Royal College of Obstetricians & Gynaecologists says that women should be informed that the additional risk of miscarriage following amniocentesis is around 1%.

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 have a termination. If you decide to have a termination following amniocentesis, this time difference can mean you need to have different type of termination due to the potentially later stage of the pregnancy.

For more information about termination of pregnancy, see Antenatal Results and Choices:
www.arc-uk.org/for-parents/ending-a-pregnancy

For more information about amniocentesis, see:
➢ NHS website:
www.nhs.uk/conditions/amniocentesis/
➢ Royal College of Obstetricians & Gynaecologists:
www.rcog.org.uk/en/guidelines-research-services/guidelines/gtg8/
Other Sources of Support:

➢ Antenatal Results and Choices (ARC)

ARC is a national charity helping parents and healthcare professionals through antenatal screening and its consequences. 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. ARC’s helpline is available Monday to Friday 10.00am to 5.30pm, 0845 077 2290 from a landline and 0207 713 7486 from a mobile. The website is: www.arc-uk.org

➢ Miscarriage Association

Information and support for anyone affected by miscarriage. Helpline: 01924 200 799 Monday to Friday 9am-4pm. Website: www.miscarriageassociation.org.uk

➢ SMA UK

SMA UK provides free, confidential information and support to anyone affected by SMA. We don’t offer a counselling service and aren’t experts in terms of future pregnancy options but we are always willing to listen and offer emotional support.

Phone: 01789 267 520

Email: supportservices@smauk.org.uk

Website: www.smauk.org.uk

We have a number of guides and resources about SMA available to download from our website or hard copies can be requested by phoning or e-mailing the support services team.
Our thanks to Alison Lashwood, Consultant Genetic Counsellor at Guys and St. Thomas’ Hospital, London for the original information on PGD. Also, to Dr Stephanie Allen and Julie Hewitt from the West Midlands Regional Genetics Laboratory for the information on NIPD.

We are grateful to all the writers and reviewers who assist us in our information production. A list of who this includes may be viewed on our website: www.smauk.org.uk/our-writers-and-reviewers-panel or requested from supportservices@smauk.org.uk

Whilst every effort is made to ensure that the information in this publication is complete, correct and up to date, this cannot be guaranteed. The collaborating organisations shall not be liable whatsoever for any damages incurred as a result of its use.

If you have any feedback about this information, please do let us know:
supportservices@smauk.org.uk