UNIVERSITY OF OXFORD

DEPARTMENT OF PAEDIATRICS
Level 3, Academic Centre, John Radcliffe, Oxford OX3 9DU
Tel: +44(0)1865 226956
www.paediatrics.ox.ac.uk

NHS Royal Berkshire NHS Foundation Trust

Population-based New-Born Screening of Spinal Muscular Atrophy to evaluate the uptake and feasibility in the UK context (NBS for SMA)

Participant Information Sheet

Dear Parents,

You have been invited to join our study on newborn screening for Spinal Muscular Atrophy (SMA). This study is completely voluntary and you can choose whether to take part or not.

During their first week, all newborns are included in the national newborn screening program. This is when a few drops of blood are taken from the newborn's heel and collected onto a blood spot card (Guthrie card) and checked for specific treatable metabolic and hormonal disorders. If you take part in our study, no extra blood will be taken and we will use the Blood spot Card to screen your newborn for SMA.

Please take your time to read the information. If you have any questions, or would like to know any more about the study, please do not hesitate to ask your research midwife who has approached you about the study. You can also contact the study team by phone or email; their contact details are at the end of this booklet. Please talk about it with your partner, family or friends, if that would help you.

Why have I been invited to this study?

We are inviting women who are more than 18 weeks pregnant or have given birth in the last 28 days, and are living in an eligible area to take part in this study with their newborn(s).

Recruitment for the study is closing on 31st December 2024 and all babies will need to be born on or before this date to be included.

Why is the study being carried out and you have been asked to take part?

Spinal Muscular Atrophy (SMA) is a rare, but treatable, genetic disease affecting approximately 1 in 10,000 births and typically presents in infancy and early childhood. SMA is caused when part of the gene important to maintain nerve cells, called survival motor neuron 1 (*SMN1*) is found to be missing (deletions) or disrupted (mutations). SMA progressively, and irreversibly, destroys the nerve cells in the brain and spinal cord, that control movement and in turn, leads to the deterioration or loss of muscle strength. Children with the most common and severe type of SMA, if left untreated, may not survive beyond 2 years of age without permanent ventilatory support and children who do, may not achieve independent sitting with even fewer ever walking independently.

There are treatments for SMA approved by UK regulatory bodies that can be given by the NHS and so far are shown to be more effective the sooner the treatment is given. This highlights the importance of screening and early detection of SMA in newborns.

The study aims to make it possible to detect SMA within the first few weeks of birth and before symptoms start to develop. The findings will help the National Screening Committee to decide whether to include SMA screening as part of the UK newborn screening programme.

What do I need to do if I decide to take part in this study?

If you would like to take part in this study, we will ask you to sign a consent form. We will also ask you to provide us with the following information:

- your name, contact details and NHS number
- your postcode
- your baby's expected or actual date of birth
- whether your pregnancy/birth is single or multiple (and if you are having twins or triplets, which baby/babies you give your consent for)

The study team will store this information securely in their database.

What will happen to my baby's blood spot card (Guthrie card)?

If you decide to take part, a portion from your baby's blood spot card will be collected and sent to the NHS Oxford Regional Genetics Laboratories, and submitted for genetic testing for SMA. The data from this sample will be securely stored for 12 months and any leftover sample will be stored and destroyed after 3 months.

What will happen if my baby's screening result is normal (negative for SMA)?

You will be emailed separately to your standard newborn screening results (via the email address you give if you consent to take part in the study) and told your baby's screening result is 'negative'. This is normal and means SMA is not suspected. The SMA test result may take longer than your standard newborn screening results. Please ensure you enter a current, personal email address on the consent form, and one that will be monitored regularly for the next 6 months to ensure you receive the results.

What happens if my baby's screening result is positive for SMA?

If your baby is found to be screened positive for SMA, you will be contacted by phone as soon as possible. You will be invited to a meeting in person with a senior consultant who is an SMA specialist and their clinical team within 2 working days, or as soon as it is practically possible for you. A second test will be done to confirm the diagnosis and here the next steps will be discussed. Any treatment that is required for your baby would be provided through the NHS and is not part of this research study.

What should I consider?

95% of people having SMA will be detected by this test- as this test looks for a specific deletion that is present in 95% of affected individuals. In 5% of people with SMA, the *SMN1* gene is mutated and that cannot be found by this screening test. If your child has the rarer mutation, we may not detect it.

In some cases, other personal details including, the baby's NHS number will be required and used by the study team for processing purposes i.e. to match the blood spot card with the consent. The postcode is to ensure that you are not out of the eligible area for the study. If after birth you have moved, it may not be possible to screen your baby for SMA.

Occasionally, due to factors outside of our control, your baby's sample might be sent to another laboratory depending on where you receive your post-natal care. This means a different laboratory processes the bloodspot card, and this, unfortunately, means we would not be able to provide you with a result.

What will happen to my and my baby's data?

The data that we obtain from you and your baby will be kept strictly confidential.

The data protection regulation requires us to tell you how we will process information about you and your baby and what the legal basis for processing is. We will be processing your data for research in the public interest. The University of Oxford is the sponsor for this study, based in the United Kingdom, and is the data controller for the NBS SMA study, therefore they are responsible for making sure all personal information from you and your baby is managed appropriately.

The NBS SMA study team, as researchers, will use information collected from you and your newborn to undertake the study, but will only use the minimum personally-identifiable information needed to do so. The information you provide us about yourself and your newborn, and the genetic information about SMA from your newborn's blood test, will be assigned a unique study code for our study database. Access to the database will be protected by a password and via a secure, encrypted connection. Responsible members of the University of Oxford and the relevant NHS Trust, may be given access to the data for monitoring and/or audit of studies to ensure that the research is complying with applicable regulations. We will keep identifiable information about you for 12 months after the study has finished. This excludes any research documents with personal information, such as consent forms, which will be held securely at the University of Oxford for 3 years after the end of the study.

If you agree to your details being held to be contacted regarding future research, we will retain a copy of your consent form securely until such time as your details are removed from our database. We will keep the consent form and your details separate from one another and any research data.

The data protection regulation provides you with control over your personal data and how it is used. However, when you agree to your information being used in research, some of those rights may be limited. You can find out more about how we use your data by contacting the study team who will be happy to answer your questions. Our contact details are at the end of this information booklet. Further information about your rights with respect to your personal data is available at https://compliance.web.ox.ac.uk/individual-rights.

What will happen to the results of this study?

The results of this study may be presented to companies, and at conferences and/or used in manuscripts, abstracts, press releases, and any other publications. Neither you or your newborn will be identifiable from any report or publication placed in the public domain.

What if I decide to withdraw from the study?

You are free to withdraw at any time. You do not need to provide a reason, and this will not affect you or your baby's medical care. If you would like to withdraw from the study, please contact the study team using the contact details at the end of this Information Sheet.

If you decide to withdraw after your newborn's bloodspot sample has been taken, but not yet processed, the study sample will be destroyed and recorded as 'taken but not processed' for the study purposes. If you decide to withdraw during the sample processing, in the rare event of a positive result we would still notify the SMA consultant/clinical care team at your nearest participating site.

What if there is a problem?

If you have any concerns about the study that you would like to discuss, or wish to complain about any aspect of the way in which you have been approached or treated during the course of this study, you should contact the study team, or you may contact the University of Oxford Research Governance, Ethics & Assurance team (RGEA) at rgea.complaints@admin.ox.ac.uk or on 01865 616480.

The University of Oxford, as Sponsor, has appropriate insurance in place in the unlikely event that you suffer any harm as a direct consequence of your participation in this study. NHS indemnity operates in respect of the clinical treatment provided.

Who is organising and funding the study?

The University of Oxford is the Sponsor for this study and it is being led at the University of Oxford, Department of Paediatrics, by Prof. Laurent Servais. The study has been funded by Novartis Gene Therapies, Roche Products Ltd., and the Oxford Biomedical Research Centre. Other than direct costs, including some midwives' salaries, none of the recruitment sites are receiving additional payment for their participation in this study.

Participation in future research

We would like to invite you to take part in future research studies for neuromuscular conditions for which your baby may be suitable. If you agree, we will retain your details in a register, held securely in the Department of Paediatrics, University of Oxford and only accessible by authorised individuals of the research team. Agreeing to be contacted does not mean you or your child has to take part in future research, and you can be removed from this register at any time you wish by contacting the Central SMA team (details below).

Who has reviewed the study?

This study has been reviewed and approved by Research Ethics Committee (REC 21/SC/0394).

Contact us

You can scan the QR codes below to register your interest and a member of the delegated SMA Research Team will contact you.

[Please scan this code to register your interest]

[Sibling's video]

Local NBS SMA Team:
Women's and Children's Research Team
Royal Berkshire NHS Foundation Trust
Research Office, Buscot Ward
Berkshire RG1 5AN
Telephone: 0118 322 8652
Email:
child.research@royalberkshire.nhs.uk

Central NBS SMA Team:
Level 3, Academic Centre
STRONG Group Offices, John Radcliffe Hospital
Oxford OX3 9DU
Telephone: 01865 618799
Email:
sma.newbornscreening@paediatrics.ox.ac.uk

Thank you for reading this information and thank you for considering taking part!

A sub-study into the acceptability of being approached to participate in the SMA NBS study is being conducted in parallel by Warwick University, details of which are below.

University of Warwick Acceptability Study: Survey of parent views on screening for Spinal Muscular Atrophy in newborn babies

If you wish to take part in the sub-study you will be provided with a separate Participant Information Sheet by Warwick. Views are being gathered from:

1) parents who have **agreed to have their baby screened** for SMA
2) parents who have **decided not to have their baby screened.**

Your views are very important as they will form part of the evidence presented to the UK National Screening Committee (UK NSC), to help them decide whether SMA should be included in routine newborn screening tests. The UK NSC would like to know more about what influences parents' decisions on whether to have screening for their child or not.

The University of Warwick will email you with more details about their study, which involves completing a short survey (5-10 min). There is also an option to speak with the Warwick team if you would be interested in doing an interview. Agreeing to be sent information about the Acceptability Study does **not** mean that you have to take part, and you can withdraw your consent to be contacted by the University of Warwick at any time.

You can also contact the Warwick team directly (see below) if you would be interested in taking part in the Acceptability Study (this includes parents who have decided not to take part in the newborn screening study for SMA).

Whether you consent to us passing on your details to Warwick or not, has no effect on your participation in our the newborn screening study for SMA and will not affect any care you or your baby receive.

Who do I contact if I have any questions or concerns about this sub - study?

For any questions or concerns about the Acceptability study (surveys or interviews), please contact us at SMAscreening@warwick.uk.

QR code to Warwick University Acceptability Study web page:

Thank you for reading this information and thank you for considering taking part!