DEPARTMENT OF PAEDIATRICS

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

Participant Information Booklet

Dear Parents,

We would like to invite you to consider taking part in our research study on newborn genetic screening for spinal muscular atrophy (SMA). Taking part is voluntary; it is entirely up to you to decide whether this is something you would like to do.

To help you decide, we have prepared this information sheet which explains why we want to do this research study and, most importantly, what taking part would involve for you and your newborn baby.

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 chosen for this study?

All babies that are born in the Thames Valley or in the Wessex region are eligible to take part in this study which tests for a condition called Spinal Muscular Atrophy (SMA). This is a rare disease, and so the vast majority of babies who are tested will not have the condition, but for those that do, early diagnosis and treatment are critical.

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The standard newborn screening programme

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A challenging aspect of treating SMA is that the diagnosis is often only made once the child has serious clinical symptoms, such as movement problems, by which point many motor nerves will have been lost. There are now some new treatments for SMA that are being assessed by the National Institute for Health and Care Excellence (NICE). The treatments have been shown to benefit children with certain types of SMA who are treated at an early stage. The treatments have been approved by UK regulatory bodies, and can be given on the NHS.

Our newborn screening study aims to make it possible to detect SMA within days of birth, before symptoms develop, so that any affected newborn can receive diagnosis and treatment at the earliest possible opportunity. We hope that the findings from this study will help the National Screening Committee decide whether SMA screening should be included in the UK newborn

taken in the first week after birth. No additional blood samples will be required. If you decide to take part, a sample will be collected from the NHS Screening Laboratory either at the John Radcliffe Hospital or at the Portsmouth Hospital which carries out all of the newborn and antenatal screening tests in the Thames Valley and Wessex regions respectively. This sample will undergo DNA extraction at the Churchill Hospital. The SMA test is a genetic test, or DNA test. The sample and data will be securely stored for 12 months and any leftover sample will be securely destroyed once the genetic analysis is complete.

Although DNA can never be completely anonymous because it is unique to each person, we will de- lp keep you and

authorised researchers at the NHS Oxford Regional Genetics Laboratory at the Churchill Hospital, based at the Oxford University Hospitals NHS Foundation Trust. The sample will not leave this laboratory.

What tests will be performed on my

The bloodspot from the Guthrie card will be collected and sent on to be received at the NHS Oxford Regional Genetics Laboratories, and submitted for genetic testing. DNA will be extracted from the sample and run on a sensitive assay called qPCR, which will allow us to detect the presence of the affected gene in SMA, called SMN1. This test is being used in many countries, and has been validated for use in the UK.

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(deletion). The genetic screening test for SMA allows us to detect this common deletion. In 5% of people with SMA, there is a rarer genetic deletion that cannot be found by the screening test. If your child has the rare deletion we may not detect it by screening.

The genetic tests used in this screening study will only detect whether your newborn will develop SMA. They will not provide any information about the likelihood that your baby will pass on SMA to their children.

be kept confidential?

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

The data protection regulation requires that we 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 SMA NBS study, therefore is responsible for making sure all personal information from you and your baby is managed appropriately.

The SMA NBS 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 to us about yourself and your

assigned a unique study code for our study database. Access to the database will also be protected by password and via a secure, encrypted connection. No data from you, or your newborn collected for this study, will leave the University of Oxford or the Oxford University Hospitals NHS Trust. Responsible members of the University of Oxford and the relevant NHS Trust, may be given access to 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.

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We would like to gather the views of parents / expectant parents who have been approached to take part in the screening study. This includes:

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

Why are we doing this study?

Your experience will help us to understand how parents decide whether to have their baby screened or not. We would like to know what influenced your decision, what were the most important issues for you, and was there anything that could have helped you with making your

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