

SPINAL MUSCULAR ATROPHY SCREENING SURVEY (UK)

A survey of the views of families living with SMA in
the UK on the possibility of population screening
for SMA

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SMA Screening Survey (UK)

Welcome to the SMA Screening Survey (UK). This survey is part of a research project called 'Imagining Futures: The Social and Ethical Implications of Genetic Screening' (http://www.warwick.ac.uk/imagining_futures). This survey has been designed to explore the views of people living with SMA (or with SMA in their family) towards the possibility of screening for SMA. 'Screening' refers to the identification of SMA (those with the condition and/or carriers) in the whole UK population, not just within families already known to be affected by the condition. A screening programme would mean that people with no history of SMA in their family would be offered the chance to undergo genetic testing for the condition.

Why is the study being done? In December 2013, after a national consultation, the UK National Screening Committee (the organisation that advises the government on issues related to screening) concluded that screening for SMA should not be offered at this time (<http://www.screening.nhs.uk/sma>). It was noted in their consultation report that very little is known about how families currently living with SMA would feel about a national screening programme for SMA. This survey is designed to address this gap in understanding, and the outcome of the survey will be submitted to the UK National Screening Committee in their next review of SMA screening policy (around 2016). In order to develop this survey, 36 in-depth interviews with people who either have SMA themselves, or have SMA in their family, were conducted to get a picture of the sorts of views people have on screening. The wide range of views that emerged from these interviews have been incorporated into the survey.

Why am I being asked to take part? You are being invited to complete this survey if you are aged over 18, live in the UK and have any form of SMA in your family (including its variant forms, e.g. SMARD, Kennedy's, Spinal Bulbar Muscular Atrophy etc.) or have the condition yourself. I am interested to hear from a range of family members (e.g. aunts/uncles, cousins, siblings, grand-parents, step- and half-relatives) or anyone who considers themselves a 'family member' of someone with SMA, regardless of whether you are biologically related, and regardless of whether your relative with SMA is still living, or has died.

How do I participate? Simply fill in the SMA Screening Survey (UK) and return it in the pre-paid envelope, or to the address found at the back of this survey. The survey takes about 15-20 minutes to complete. Please complete the survey *as an individual*, rather than as a couple or family, as everyone feels differently about screening (even within couples and families) and it is important to get an accurate picture of the range of views on this topic. If you prefer, you may complete this survey online at: <http://www.warwick.ac.uk/smascreeningsurvey>

What will happen to my information if I participate? The anonymised summary results of this survey (not individual data) will be published in academic journals, written up as a research report for SMA Support UK's (formerly the Jennifer Trust) 'Inspirations' newsletter, as well as the UK SMA Patient Registry newsletter and used for conference presentations (academic, professional and patient). It will also be submitted as research evidence to the UK National Screening Committee in their next review of the policy on screening for SMA in the UK. Upon completion of the Imagining Futures research project, the anonymised data from this survey will be archived with the Economic and Social Data Service (<https://www.esds.ac.uk/>) so that in the future, other researchers may make use of the data. ***If you do not want your anonymised survey answers to be archived, please contact us (SMAscreeningsurvey@warwick.ac.uk) BEFORE completing the survey.***

What if I change my mind? If you start the survey and decide you do not want to continue, simply do not return it. However, once a completed survey is returned, it will not be possible to withdraw it from the study. This is because the survey is anonymous and therefore it will be impossible to link an individual back to their survey and remove it from the study.

Will my taking part be kept confidential? All data collected from this survey will be held anonymously and securely using data encryption software. No data which may identify you (e.g. your name/address) will be asked for, but you will be asked for background information about yourself, as well as the type of SMA affecting your family. This is in order that we can get a clearer understanding of the backgrounds of the people responding. All data will be handled in strict accordance with the Data Protection Act 1998.

Who is conducting the research? This research is being conducted by Dr. Felicity Boardman, at Warwick Medical School, and is funded by the Economic and Social Research Council. The research was given a favourable opinion by Warwick's Biomedical and Scientific Research Ethics Committee on 15/7/14 (REF: REGO-2014-903).

Are there any disadvantages to taking part? Some people may find the topic of screening for SMA distressing. Should the completion of this survey raise any issues for you which you would like support with, or further information on, please contact SMA Support UK (formerly the Jennifer Trust for Spinal Muscular Atrophy) www.smasupportuk.org.uk/ (01789 267520).

What if there's a problem? Any complaint about the way you have been dealt with during this study, or any harm you may have suffered, will be properly addressed. Please address your complaint to: Jo Horsburgh, Deputy Registrar, Deputy Registrar's Office, University of Warwick, Coventry, CV4 8UW. J.Horsburgh@warwick.ac.uk

If you have any comments or queries about the survey, or to request further paper copies, please contact us at SMAscreeningsurvey@warwick.ac.uk or complete the comments box at the end of the survey. ***You may want to tear off and keep the cover page of this survey before returning it in order to retain the project details. Alternatively, email the SMA Screening Resource and ask for a copy of this information to be posted or emailed to you.***

Thank you!

SMA Screening Survey (UK)

Section 1: About You

In this section you will be asked for basic background information about yourself. These questions are being asked in order that the researcher can gain a better understanding of the social backgrounds of people living with SMA in their family.

1. What is your sex?

- Male Female

2. What is your age?

- 18-25 years
 26-34 years
 35-45 years
 46-55 years
 56-65 years
 66+ years

3. What is your highest qualification level?

- No qualifications
 GCSE or O Level
 GCE, A level or similar
 Vocational (BTEC/NVQ/Diploma)
 Degree level or above
 Other (please specify): _____

4. What is your ethnic group? (Continued over the page)

- White- British
 White- Irish
 White- Gypsy or Traveller
 White- European
 Mixed- White and Black Caribbean
 Mixed- White and Black African
 Mixed- White and Asian
 Asian- Indian
 Asian- Pakistani
 Asian- Bangladeshi
 Asian- Chinese
 Black- African
 Black- Caribbean
 Arab
 Prefer not to say
 Other (please specify): _____

5. Do you have a religious faith?

Yes No Prefer not to say

If yes, how would you describe your religious faith?

Christian (any denomination)

Jewish

Muslim

Sikh

Hindu

Buddhist

Other (please specify): _____

Section 2: SMA and Your Family

In this section you will be asked questions about the number of people affected by SMA in your family, the type of SMA in your family as well as your perceptions of your own, and your family members' health and well-being

6. Do you have any children? (This might include step-children, foster children, adopted children or any child for whom you consider yourself to be the parent or legal guardian)

Yes No

If yes, how many children do you have? _____

7. What is your relationship to SMA? (please tick all that apply)

I have SMA myself Someone in my family has, or has died from, SMA

8. What type of SMA do you have yourself, or do you have in your family? (please tick all that apply)

Don't Know

Type 0 (in utero onset)

Type I (also known as Werdnig-Hoffman Disease)

Type II

Type III (also known as Kugelberg-Welander Disease)

Type IV

Distal Spinal Muscular Atrophy

Spinal Muscular Atrophy and Respiratory Distress (SMARD)

Other (please specify): _____

9. Please list your relationships to family members (to a maximum of 10) affected by SMA and their type of SMA (if known). This may include extended family members. If your family member(s) have died, please state their approximate year of death and age at death (if known). If no one else in your family has SMA, please go on to question 11.

	Relationship to family member	Type of SMA	Year of death (if applicable)	Age at death (if applicable)
a. Family member 1	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
b. Family member 2	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
c. Family member 3	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
d. Family member 4	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
e. Family member 5	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
f. Family member 6	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
g. Family member 7	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
h. Family member 8	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
i. Family member 9	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>
j. Family member 10	<input type="text"/>	<input type="text"/>	<input type="text"/>	<input type="text"/>

10. Do you currently, or have you at any point in the past, lived in the same household as your family member(s) listed in question 9 (please answer for each family member as listed in the order above)? 'Living in the same household' would include living together on a temporary basis with your family member(s) affected by SMA, e.g. regular holidays or stays of more than two weeks in the same household.

	Currently, or at some point in the past, lived in the same household?	
	Yes	No
a. Family member 1	<input type="radio"/>	<input type="radio"/>
b. Family member 2	<input type="radio"/>	<input type="radio"/>
c. Family member 3	<input type="radio"/>	<input type="radio"/>
d. Family member 4	<input type="radio"/>	<input type="radio"/>
e. Family member 5	<input type="radio"/>	<input type="radio"/>
f. Family member 6	<input type="radio"/>	<input type="radio"/>
g. Family member 7	<input type="radio"/>	<input type="radio"/>
h. Family member 8	<input type="radio"/>	<input type="radio"/>
i. Family member 9	<input type="radio"/>	<input type="radio"/>
j. Family member 10	<input type="radio"/>	<input type="radio"/>

11. How would you rate your current health and well-being?

- Very good Good Fair Bad Very Bad

12. How would you rate the current health and well-being of your family members affected by SMA listed in question 9? Please rate in the order you listed them in above. If you have no family members affected by SMA, please go on to Section 3.

	Current Health and Well-being of Family Members with SMA						
	Very good	Good	Fair	Bad	Very Bad	Don't know	Not applicable (family member died)
a. Family member 1	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
b. Family member 2	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
c. Family member 3	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
d. Family member 4	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
e. Family member 5	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
f. Family member 6	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
g. Family member 7	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
h. Family member 8	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
i. Family member 9	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
j. Family member 10	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Section 3: Your Use of Genetic and Screening Technologies

In this section, you will be asked about your previous use of genetic and screening technologies for SMA and other conditions.

13. Are you, or your partner, currently pregnant or trying to get pregnant?

- Yes
 No
 Prefer not to say

14. In your current, or previous pregnancies, have you, or your partner, ever used any of the following technologies? Please tick all that apply. Please see 'Glossary' at the end of the survey for further explanation of these technologies. (Please go on to Section 4 if you, or your partner, have never been pregnant before)

- CVS or Amniocentesis (diagnostic testing in pregnancy) for SMA
 CVS or Amniocentesis (diagnostic testing in pregnancy) for a condition other than SMA
 Pre-Implantation Genetic Diagnosis (PGD) (creation of embryos using IVF prior to testing) for SMA
 Pre-Implantation Genetic Diagnosis (PGD) (creation of embryos using IVF prior to testing) for a condition other than SMA
 Screening for Down's Syndrome (usually offered as a scan and/or blood test at around 12 weeks of pregnancy)
 Carrier Testing for SMA (a test to see if you, or your partner, are a carrier of SMA)
 None of the above

15. Have you, or your partner, ever undergone a pregnancy termination (abortion) due to a prenatal diagnosis of SMA?

Yes No Prefer not to say

16. Have you, or your partner, ever undergone a pregnancy termination (abortion) due to a prenatal diagnosis of a condition other than SMA?

Yes No Prefer not to say

16 a. If yes, please state for which condition(s) _____

Section 4: Your Views About SMA

In this section, you will be asked about your views about SMA as a condition. You will be asked about how far you agree, or disagree, with, a list of statements. These statements were derived from interviews conducted with families living with SMA and represent a wide range of views on the condition. This survey will measure how widespread these views are and how strongly they are held by the wider population of families living with SMA. Please note, there are no right and wrong answers to these questions and everyone feels differently.

17. Please state how far you agree, or disagree with, the following statements about SMA.

	Views About SMA					
	Strongly agree	Agree	Neither agree nor disagree	Disagree	Strongly disagree	Don't know
a. People with SMA can have a good quality of life	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
b. Having SMA causes people to suffer	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
c. People with SMA have heightened intelligence	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
d. People with SMA and their families are well supported by society	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>
e. Quality of life varies greatly across the types of SMA	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>

Section 5: Your Views on Screening for SMA

In this section, you will be asked your views on screening for SMA. You will be asked how far you agree, or disagree with a list of statements about SMA, derived from interviews with families living with SMA. These statements represent a wide range of views. This survey will measure how widespread these views are, and how strongly they are held by the wider population of families living with SMA.

Screening for SMA would involve the identification of SMA in the whole UK population, not just within families already known to be affected by the condition. However, screening in the general population could not accurately diagnose the type of SMA in that family.

Screening for SMA could be done in different ways:

1) **Pre-conception genetic screening** would identify 'carriers' of SMA (people who carry the faulty gene associated with SMA but do not have symptoms of SMA themselves) before they have children, or whilst

Thank you!

Thank you for completing the SMA Screening Survey (UK). Please return your survey in the pre-paid envelope, or to the following address:

**SMA Screening Survey (UK)
C/O Dr. Felicity Boardman,
Division of Health Sciences,
Warwick Medical School
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Coventry
CV4 7AL**

Some people may find the topic of screening distressing. Should the completion of this survey raise any issues for you which you would like to discuss further, please contact SMA Support UK (formerly the Jennifer Trust for SMA) www.smasupportuk.org.uk/ (01789 267520).

Please feel free to pass this survey on to your relatives, and any friends/acquaintances you know with SMA in their family, however please be aware that screening can be a sensitive topic, and the views of others may not be the same as yours. You can contact us for extra paper copies (SMAscreeningsurvey@warwick.ac.uk) or use the link to the online version: <http://www.warwick.ac.uk/smascreeningsurvey>

You can find out more about the Imagining Futures research project by visiting: www.warwick.ac.uk/imagining_futures

Glossary of Terms

CVS/Amniocentesis- CVS and amniocentesis are diagnostic procedures used in pregnancy which usually involve the insertion of a needle through the abdomen to remove samples to be genetically tested.

Pre-Implantation Genetic Diagnosis- refers to the creation of embryos using IVF procedures. The embryos can then be tested for genetic conditions, such as SMA, before being transferred back to the mother's uterus.

Screening for Down's Syndrome- Down's Syndrome is a chromosomal disorder caused by an extra copy of chromosome 21 which leads to varying degrees of learning difficulty. It is also known to be associated with particular health problems, including heart problems, reduced vision and hearing, as well as early-onset Alzheimer's Disease (a form of senile Dementia). Screening for Down's Syndrome is offered to all pregnant women in the UK and usually involves a blood test and/or a scan (to measure the foetus' nuchal fold- the back of the neck) at around 12 weeks of pregnancy.

If you have any comments about the SMA Screening Survey (UK) please write them in the box below: