Study of Adults and Adolescents with Russell Silver syndrome in the UK (STAARS UK)

INFORMATION BOOKLET

For participants and parents/guardians

Contact the research team to take part in this study

REC reference number: 13/SC/0630
Version 2 Date: 03/01/14

Part 1
Invitation

This is a study to find out more about what it means to have Russell Silver syndrome (RSS), a genetic condition affecting growth. We want to understand why it happens and whether there are long term health issues.

Any person aged 13 years or more who has been diagnosed with RSS can take part. If a diagnostic test for RSS has not yet been offered, we will help to arrange this.

Before deciding to take part, it is important to understand why the research is being done and what it will involve. Please take the time to read this carefully and discuss it with anyone you wish. Ask us if there is anything that is not clear or if you would like more information.

The study is trying to understand more about the development of people with Russell Silver syndrome as they get older. We are interested in growth, how the body is made up, and how the body uses energy (metabolism) in general. We are also finding out more about the major issues around living with Russell Silver syndrome. The study will look at the genetic cause and try to work out why it happened. One of the main reasons for the study happening now is that we are trying to find out whether the use of growth hormone has had an impact and so we want to include people who have and have not had this treatment. Participation in the study involves coming to a clinic at one of several hospital outpatient research centres across the UK for a discussion about medical history, an examination and some tests. The visit will last around 3 hours.

Taking part in this study is voluntary and does not impact on standard NHS care.

What are the possible benefits of taking part?

The main benefit of participating is that the study may help to improve the understanding of growth, development and health issues of people with Russell Silver syndrome; in the hope of improving knowledge so that appropriate treatment might be offered to people with this condition.

The study visit will be a one-to-one appointment with a clinician where a full health check will be performed over a longer time than is usually available at a healthcare appointment. By the end of the visit, any participant will hopefully have learnt more about his or her body and health, and discussed issues with a team who are meeting many people with this rare condition all the time. There will be the option to be given test results such as blood pressure and blood tests. If there are any problems identified with general health during the assessment, an appropriate referral can be made, with your permission.

Some people have been diagnosed with Russell Silver syndrome based on clinical assessment. They might not have been able to have a genetic test before or may have been tested when fewer genetic changes were known. Another benefit of this study is that those participants will be able to have the current tests for the known genetic changes.

What will happen at the research appointment and what measurements will be taken?

In brief

The study will involve talking to a research clinician, being examined, and having photos and a blood test taken. Because of the nature of the blood tests we need all participants to come to the clinic having fasted overnight (or for at least 6 hours). We will start with the blood sample followed by a snack before the rest of the interview.

In addition, we will ask everyone for permission to make contact about another part of the study, which involves a one-to-one interview about what it is like to have RSS.

Medical interview and examination

We will ask about medical, birth and family details, including information about early childhood, growth as a child, and any medication taken. There will be some questions about lifestyle using a short questionnaire.

If parents come to the study clinic, there is a questionnaire to complete; if not, we can post it where possible. This questionnaire asks about the conception, pregnancy, early medical problems, parent health and family health.

If agreed, we will perform a full medical examination including blood pressure and growth measurements such as height, weight, and head size. To check how well their lungs work, participants will be asked to blow into a lung machine. We will take measurements of skin thickness and grip strength, which tell us about muscle and fat. Passing a tiny electrical current between sensors stuck on the skin helps us work out body structure. This is tolerated well in many studies as it does not hurt and people only feel the stickers.

We will ask for permission for the research clinician to take photographs (of the face and full length, while standing fully clothed). These will help us to show how people with RSS look at different ages. The photos may be included when we write reports but only with agreement. It is important to let you know that nowadays reports are also available online so these photos would be on the internet but names would not be added.

Scans: if the study visit is at a centre where scans are available, we will measure the density of different bones. There are two different machines and they are painless. The DEXA scan measures bone density at the hip and spine whereas the pQCT scan measures the density in the arm and leg. To do both scans takes about 30 minutes. They have been used in many studies in children and adults before and are safe. They involve a very low dose of radiation – it is about the same as 3 days background radiation or the same as a chest x-ray. This will not have any implication for the participant's health and it is optional.

Blood: we would like to take a small sample of blood, about 2 tablespoons. We can use a cream or spray to numb the arm if wanted and then take a small sample with a needle. This procedure has been used for taking blood samples in many other studies and usually people cope well. The results will give us information on body chemicals and nutrients in the blood so that we can understand energy handling in the body.

Genetic testing: we would like to find out the genetic cause of Russell Silver syndrome. The test will involve taking a painless swab from the inside of the cheek. We will be looking for genes that might cause RSS using techniques that look at large parts of the genetic material. We may work with other scientists involved in RSS research. That could mean part of the sample being sent to other centres outside the UK. In that case the medical information would stay with the research team in Southampton and the sample would only be identified using the study number.

Hair and skin samples: at the study visit, we will ask participants aged 18 years and over if we may also take a hair sample and a skin sample. This will allow us to grow more cells in the laboratory - called a "cell line" - and allow us to use the cells without having to request any further blood samples. We will be able to keep these cells growing indefinitely and use them for future research purposes. They may be used in ways we have not yet planned. These will not be labelled with a name. A participant does not need to have this test to join the study.

Examination, blood sample and genetics results

If something out of the ordinary is identified at the examination we will discuss it and, with permission, contact the appropriate GP if needed.

Most of the test results will not be provided as they will be performed together and so will be out of date by the time the results are known. But some results can be requested - like cholesterol, fasting blood sugar, body mass index, and (where available) bone density. Genetic diagnostic test results will be provided for participants on request or if they differ from diagnostic results already provided by the participant. The research is not designed to report any unexpected genetic findings unrelated to RSS.

We will send regular information updates on the overall results and progress of the study.

In-depth interview

Participants will be asked if they would take part in another in depth discussion at another time, which could take around 2 hours. This is to try and discover more about what it means to live with Russell Silver syndrome. All participants will be asked if they would like to participate further, but only some people will actually be interviewed so we get a broad spread of ages.

Expenses and payments

Your travel expenses will be reimbursed at rates recommended by the University of Southampton.

What if there is a problem?

Any complaint about the study or any possible harm will be addressed. Detailed information on this is given in Part 2.

Is taking part in the study confidential?

As always, all information about participation in this study will be kept confidential. Details are included in Part 2.

Contact Details:

To contact the study team at any time phone:
02381206551

This completes Part 1 of the Information Sheet. Please read Part 2 before making a decision.

Part 2

What if there is a problem?

If you are worried about any aspect of this study, please speak to the researchers who will do their best to answer your questions.

Under our formal research procedures we are required to give you the following information:

If you remain unhappy and wish to complain formally, you can do this through the NHS Complaints Procedure. Details can be obtained from the study coordinator. We are an experienced research team, and aim never to cause you harm. As outlined in Part 1, the planned investigations are considered safe. In the very unlikely event that something does go wrong and you/your child are harmed due to someone's negligence then you may have grounds for a legal action for compensation against the University Hospital Southampton but you may have to pay your legal costs.

Will our taking part in this study be kept confidential?

All information collected about you and your family during the course of the research will be kept strictly confidential. If we discover information that may be useful for your family doctor with your permission we will contact your doctor.

With your permission, the research team will have access to your medical notes. From time to time monitors and auditors from NHS Research and Development offices and regulatory inspectors may also require access to your clinical notes to check data from the study.

We will keep the information collected in the study for 15 years in line with research guidance for University Hospital Southampton. After this time it will be destroyed.

What will happen to the results of the research study?

The results of the study will be published in medical journals so that doctors and health professionals all over the world can understand what increases the likelihood of illness in child and adulthood with

RSS. We will also arrange for parent groups such as the Child Growth Foundation to write about the study results so that we can tell others what we have found.

We have a website that is kept updated with the findings from this study: http://www.southampton.ac.uk/geneticimprinting/xxxxxx

Who is organising and funding the research?

This research is funded by the National Institute for Health Research (NIHR). The study is organised by the University Hospital Southampton and the University of Southampton.

Who has reviewed the study?

This study was given ethical approval by NRES Committee South Central – Hampshire B and the National Institute for Health Research.

THANK YOU FOR READING THIS BOOKLET

If you have any further questions, please feel free to ask

Contact us:

STAARS UK
Wessex Clinical Genetics Service
Princess Anne Hospital
Southampton
SO16 5YA
Tel: 02381 206551

To take part in this study either contact the health professional sending you this form or contact:

The UK Russell Silver project team on

1. Phone 023 8120 6551
2. E-mail……..
3. Return this section and send to

Study of Adults and Adolescents with Russell Silver syndrome in the UK (STAARS UK)
Wessex Clinical Genetics Service
Princess Anne Hospital
Coxford Road
Southampton
SO16 5YA

Provide us with:

NAME

ADDRESS

PREFERRED CONTACT NUMBER