PARTICIPANT INFORMATION SHEET (PIS)

Study Title:
Down Syndrome Regression Disorder (DSRD) Delphi Survey in the United Kingdom

Chief Investigator: Dr Ella Rachamim

We'd like to invite you to take part in our survey as part of a research study we are carrying out. It should only take around 5 minutes to complete. Before you decide whether or not you wish to take part, we would like to explain the background to this study, why this research is being done and what it would involve for you.

The first part of this Participant Information Sheet (PIS) explains the purpose of the study and what will happen if you take part. Part two gives you more detailed information about how the study is being conducted.

Part 1

What is the purpose of the study?

A group of children and young people with Down Syndrome (DS) have been identified to have sudden significant deterioration in their motor, language and behaviour skills, most often preceded by a potential emotional or environmental trigger, such as a change in school or family circumstances. This is named as Down Syndrome Regression Disorder (DSRD) but there have been a number of different medical terms used in the literature which can make it harder for research. Although some therapies have been tried and some are still under research, no definitive treatments have been approved. This disorder is a separate entity to dementia and to autism – however, poor awareness amongst professionals of DSRD along with some of the overlap of symptoms with autism and dementia, has meant that little is known about the true incidence and therefore management of DSRD.

The questions are aimed at identifying any potential triggers like difficult experiences and challenging issues that the young person has encountered in the past.

In the United Kingdom, the exact occurrence of this condition remains unknown. A series of symptoms and triggering factors have been developed recently, by researchers and clinicians, that are very helpful in potentially identifying this condition. The survey questions relate to these series of symptoms and triggers that could possibly help in identifying DSRD. Our vision is that by identifying the number of children and young people with DSRD in the United Kingdom, it will help to improve knowledge and awareness of this condition and in turn, will lead to research on the management of this condition, in the UK, in the future.

Why have I been invited?

All parents, carers and professionals involved in the care of children and young people with Down Syndrome who are aged between 10 and 40 years, are eligible to take part in this study. We are sending the survey to all professionals, carers and parents across the UK who may fall into this group.

Do I have to take part?

It is optional to take part in the study and your decision. If you do decide to take part in the survey, it implies you have consented for the research study. A decision to not participate or withdraw at any time or not take part will not affect the standard of care you receive.

What would taking part involve?
This is a one-off survey. If you have/ know more than one child or young person who is between 10 and 40 years of age with Down Syndrome who could be showing DSRD symptoms, you can fill the survey for separately for each person. The questionnaire is only for the purpose of identifying the occurrence of this condition and to gain knowledge on this. No further research will be extrapolated from this survey. The survey is fully anonymous. The demographic data being gathered is only to gain insights on the occurrence of DSRD.

The results of the survey will be discussed in an expert panel meeting, sharing the findings with relevant researchers and leaders in the field, and with the worldwide Down Syndrome research community. All participants will be updated on the outcome of the survey and of any future research through the same platform the survey is being circulated from.

The survey maintains confidentiality of the participants. All responses will be collected jointly by Dr Ella Rachamim (Chief Investigator and Community Paediatrician, Edgware community hospital, London) and Dr Abinaya Seenivasan (Paediatric Neurology Trainee, Royal Manchester Children's Hospital, Manchester). The data will be analysed through Microsoft excel software and will be discussed in the expert panel meeting. The demographic questions are optional and if you do not wish to declare any demographic details, it is not essential at all, and you can still proceed with the rest of the survey.

If you want to gain more information about the results of the survey and further information on the topic, you are welcome to contact a member of the survey team, through the platform that the survey has been sent to you from.

What are the possible benefits of taking part?
We are optimistic that the information findings from the survey and the awareness that the survey will bring of DSRD will help improve the treatment of people with DSRD.

What are the possible disadvantages and risks of taking part?
This is an anonymous survey, hence the potential disadvantages and risks are negligible. The survey results are gathered only by the research team and this, along with the anonymity of the survey and that we have no other collaborators involved in the data collection or sharing, ensures confidentiality.

There is an extremely small risk of breach in confidentiality due to the rare incidence of this condition and the small proportion of the study group, meaning the research team might be able to work out who the survey is describing, despite it being totally anonymous. However, in order to eliminate this risk, we will ensure that the data is processed only by the research members, who will also be the same members who deliver the results to the expert committee panel.

Following the survey, we will store the data for a period of 5 years, in case of any further research developments in the future. At this stage, we are not intending to use the data for any further research.

Whilst completing the survey on behalf of a child or young person in your care, if further questions or needs arise, we advise you to contact the professionals involved in their care for further support.

Informing General Practitioner / other healthcare practitioner

- Please make sure that the participants are between 10 and 40 years of age.
- This is an anonymous survey which is for the purpose of identifying the incidence of DSRD.

There is a professional version of the survey that your GP or other health care professionals might be answering on your behalf. We are not informing your GP of your participation and we do not require information from your clinical records for this study.

What do I have to do?
The survey will be sent as an online link to open and with a QR code that can be scanned. You can use either of those to fill-in the questions. The entire survey takes less than 5 minutes to complete, and we recommend you fill the survey in a quiet and relaxed environment.

What if relevant new information becomes available?
We plan to communicate with you about the results of the survey through the same communication team/platforms that were used to send out the survey. You will also be notified of any future research discussions.

What if there is a problem?
Any complaint about the way you have been dealt with or any possible harm you might suffer will be addressed. The detailed information on this is given in part 2.

This completes Part 1. If the information in Part 1 has interested you and you are considering participation, please read the additional information in part 2 before making any decision.

Part 2

What if there is a problem?
Complaints
If you wish to complain or have any concerns about any aspect of the way you have been treated during the course of this study then you speak with the researchers who will do their best to answer your questions or concerns; Dr Ella Rachamim, erachamim@nhs.net or Dr Abinaya Seenivasan, abinaya.seenivasan@nhs.net

The normal National Health Service complaints mechanisms are also available to you. If you are still not satisfied with the response, you may contact the Research and Development Office at the Royal Free at rf.randd@nhs.net or the Patient advice & liaison service (PALS):
Barnet Hospital PALS: 020 8216 4924
Chase Farm Hospital PALS: 020 8216 4924
Royal Free Hospital PALS: 020 7472 6446 or 020 7472 6447
Fax: 020 7472 6463
Text: 07624 803635 (deaf users only)
E-mail: rf.pals@nhs.net / bcfpals@nhs.net
The PALS office is on the ground floor in the hospital's main reception. The patient advice and liaison service for Barnet Hospital and Chase Farm Hospital is based on the ground floor of Barnet Hospital, near the main entrance. The PALS office is open Monday to Friday 10am-4pm.

What will happen to the results of the research study?
This is a national wide survey, and the results will be interpreted as broad scientific results to identify the incidence of this DSRD in the UK. Further research is required to consider any results relevant to the individual and it is not being explored in the survey.

The results of the expert committee panel will be conveyed through the same platform used to distribute the survey. The results might be published as a scientific data in international journals as a generic survey result. No patient identifiable data will be reported and the participants will not be identified in any report/publication unless they have given their consent.

Discovering health related findings
This survey is in the initial stages of identifying DSRD. The results are subjected to the expert committee opinion and consensus.

Who is organising and funding the research?
The research is being organised by the sponsor who oversees the conduct of the trial ie The Royal Free London NHS Foundation Trust. The research is an online questionnaire survey and is not being funded by any sources.

Commercial Involvement
Nil

Expenses and payments
This is an online survey and no expenses or payment to participants are applicable.

How have patients and the public been involved in this study?
DSRD has been predominantly considered due to parental and carer reports of symptoms in their child or young person, that can't be explained and that have led to a significant regression in their functioning. Symptoms relating to a deterioration from the person's baseline are debilitating and impacts on the person with Down Syndrome and everyone involved in that person's care. This is an under-explored area and not understood clearly, with poor awareness amongst professionals, but has some increasing research interest worldwide, which we hope will pave the way for more work to be done in the UK on DSRD. The survey is based exclusively on the symptomatology and the triggers around the child or young person's life. Furthermore, this will help guide us when bringing together the appropriate professionals to get involved.
Parent and carer involvement will be key, so the role of the public is pivotal.

Who has reviewed the study?
All research in the NHS is looked at by independent group of people, called a Research Ethics Committee (REC), to protect your interests. This study has been reviewed and given favorable opinion by Royal Free Research Ethics Committee.

Further Information and Contact Details
General information about research
https://www.downs-syndrome.org.uk/wp-content/uploads/2023/10/Regression-Factsheet-FINAL-27.10.23.pdf

Data Protection transparency information
This is a questionnaire survey with anonymous data collection.

Specific information about this research study
https://pubmed.ncbi.nlm.nih.gov/35911905- Assessment and Diagnosis of Down Syndrome Regression Disorder: International Expert Consensus. By Santoro JD et al, Front Neurol. 2022 Jul 15;13:940175. doi: 10.3389

Advice as to whether they should participate:
Down Syndrome Medical Interest Group (DSMIG)- https://www.dsmig.org.uk

Who they should approach if they are unhappy with the study:
Please email erachamim@nhs.net
Abinaya.seenivasan@nhs.net

Thank you for taking the time to read this information sheet. If you would like to take part, you will be asked to sign a consent form; one for you to keep, one for the investigator and one for your medical records.

Patient data and research

This leaflet explains how health research uses information from patients. If you are asked to take part in research, you can ask what will happen in the study.

What is patient data?

When you go to your GP or hospital, the doctors and others looking after you will record information about your health. This will include your health problems, and the tests and treatment you have had. They might want to know about family history, if you smoke or what work you do. All this information that is recorded about you is called patient data or patient information.

When information about your health care joins together with information that can show who you are (like your name or NHS number) it is called identifiable patient information. It's important to all of us that this identifiable patient information is kept confidential to the patient and the people who need to know relevant bits of that information to look after the patient. There are special rules to keep confidential patient information safe and secure.

What sort of patient data does health and care research use?

There are lots of different types of health and care research.

If you take part in a clinical trial, researchers will be testing a medicine or other treatment. Or you may take part in a research study where you have some health tests or answer some questions. When you

have agreed to take part in the study, the research team may look at your medical history and ask you questions to see if you are suitable for the study. During the study you may have blood tests or other health checks, and you may complete questionnaires. The research team will record this data in special forms and combine it with the information from everyone else in the study. This recorded information is research data.

In other types of research, you won't need to do anything different, but the research team will be looking at some of your health records. This sort of research may use some data from your GP, hospital or central NHS records. Some research will combine these records with information from other places, like schools or social care. The information that the researcher collects from the health records is research data.

Why does health and care research use information from patients?

In clinical trials, the researchers are collecting data that will tell them whether one treatment is better or worse than other. The information they collect will show how safe a treatment is, or whether it is making a difference to your health. Different people can respond differently to a treatment. By collecting information from lots of people, researchers can use statistics to work out what effect a treatment is having.

Other types of research will collect data from lots of health records to look for patterns. It might be looking to see if any problems happen more in patients taking a medicine. Or to see if people who have screening tests are more likely to stay healthier.

Some research will use blood tests or samples along with information about the patient's health. Researchers may be looking at changes in cells or chemicals due to a disease.

All research should only use the patient data that it really needs to do the research.
You can ask what parts of your health records will be looked at.

How does research use patient data?

If you take part in some types of research, like clinical trials, some of the research team will need to know your name and contact details so they can contact you about your research appointments, or to send you questionnaires. Researchers must always make sure that as few people as possible can see this sort of information that can show who you are.

In lots of research, most of the research team will not need to know your name. In these cases, someone will remove your name from the research data and replace it with a code number. This is called coded data, or the technical term is pseudonymised data. For example, your blood test might be labelled with your code number instead of your name. It can be matched up with the rest of the data relating to you by the code number.

In other research, only the doctor copying the data from your health records will know your name. They will replace your name with a code number. They will also make sure that any other information that could show who you are is removed. For example, instead of using your date of birth they will give the research team your age. When there is no information that could show who you are, this is called anonymous data.

Where will my data go?

Sometimes your own doctor or care team will be involved in doing a research study. Often, they will be part of a bigger research team. This may involve other hospitals, or universities or companies developing new treatments. Sometimes parts of the research team will be in other countries. You can ask about where your data will go. You can also check whether the data they get will include information that could show who you are. Research teams in other countries must stick to the rules that the UK uses.

All the computers storing patient data must meet special security arrangements.

If you want to find out more about how companies develop and sell new medicines, the Association of the British Pharmaceutical Industry has information on its website at http://www.abpi.org.uk.

What are my choices about my patient data?

- You can stop being part of a research study at any time, without giving a reason, but the research team will keep the research data about you that they already have. You can find out what would happen with your data before you agree to take part in a study.
- In some studies, once you have finished treatment the research team will continue to collect some information from your doctor or from central NHS records over a few months or years so the research team can track your health. If you do not want this to happen, you can say you want to stop any more information being collected.
- Researchers need to manage your records in specific ways for the research to be reliable. This means that they won't be able to let you see or change the data they hold about you. Research could go wrong if data is removed or changed.

What happens to my research data after the study?

Researchers must make sure they write the reports about the study in a way that no-one can work out that you took part in the study.

Once they have finished the study, the research team will keep the research data for several years, in case they need to check it. You can ask about who will keep it, whether it includes your name, and how long they will keep it.

Usually your hospital or GP where you are taking part in the study will keep a copy of the research data along with your name. The organisation running the research will usually only keep a coded copy of your research data, without your name included. This is kept so the results can be checked.

If you agree to take part in a research study, you may get the choice to give your research data from this study for future research. Sometimes this future research may use research data that has had your name and NHS number removed. Or it may use research data that could show who you are. You will be told what options there are. You will get details if your research data will be joined up with other information about you or your health, such as from your GP or social services.

Once your details like your name or NHS number have been removed, other researchers won't be able to contact you to ask you about future research.

Any information that could show who you are will be held safely with strict limits on who can access it.

You may also have the choice for the hospital or researchers to keep your contact details and some of your health information, so they can invite you to take part in future clinical trials or other studies. Your data will not be used to sell you anything. It will not be given to other organisations or companies except for research.

Will the use of my data meet GDPR rules?

GDPR stands for the General Data Protection Regulation. In the UK we follow the GDPR rules and have a law called the Data Protection Act. All research using patient data must follow UK laws and rules.

Universities, NHS organisations and companies may use patient data to do research to make health and care better.

When companies do research to develop new treatments, they need to be able to prove that they need to use patient data for the research, and that they need to do the research to develop new treatments. In legal terms this means that they have a 'legitimate interest' in using patient data.

Universities and the NHS are funded from taxes and they are expected to do research as part of their job. They still need to be able to prove that they need to use patient data for the research. In legal terms this means that they use patient data as part of 'a task in the public interest'.

If they could do the research without using patient data they would not be allowed to get your data.

Researchers must show that their research takes account of the views of patients and ordinary members of the public. They must also show how they protect the privacy of the people who take part. An NHS research ethics committee checks this before the research starts.

What if I don't want my patient data used for research?

You will have a choice about taking part in a clinical trial testing a treatment. If you choose not to take part, that is fine.

In most cases you will also have a choice about your patient data being used for other types of research. There are two cases where this might not happen:

1. When the research is using anonymous information. Because it's anonymous, the research team don't know whose data it is and can't ask you.
2. When it would not be possible for the research team to ask everyone. This would usually be because of the number of people who would have to be contacted. Sometimes it will be because the research could be biased if some people chose not to agree. In this case a special NHS group will check that the reasons are valid. You can opt-out of your data being used for this sort of research. You can ask your GP about opting-out, or you can find out more at https://www.nhs.uk/your-nhs-data-matters/.

Who can I contact if I have a complaint?

If you want to complain about how researchers have handled your information, you should contact the research team. If you are not happy after that, you can contact the Data Protection Officer:

Data Protection Officer
Kevin Winter
kevinwinter@nhs.net
020 7794 0500 (hospital switchboard)

If you are not happy with their response or believe they are processing your data in a way that is not right or lawful, you can complain to the Information Commissioner's Office (ICO) (www.ico.org.uk or 0303 123 1113).