



**prisms**  
**2022**

*Envisioning  
the Possibilities*

# Smith-Magenis Syndrome Patient Registry

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## Presentation Topics

- Patient Registry
- What is asked
- Let's Talk About the Data
- Ready to Participate

# Patient Registry Topics

What is a patient registry?



Why is a patient registry needed?



What is needed to participate in the patient registry?



Who is eligible to participate in the patient registry?

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# What is a patient registry?

- A large database
- Used to collect and store large amounts of information
- Over a long period of time
- This registry is an online tool to collect and store medically relevant information on individuals with Smith-Magenis syndrome

# Why is a patient registry needed?

- Allows researchers to better understand the natural history of Smith-Magenis syndrome (SMS)
- Allows for the collection of similar information in one streamlined way enabling researchers to work collaboratively to create a better understanding of SMS
- Improved understanding of SMS can lead to better care for individuals with SMS

# Who is eligible to participate in the patient registry?

- ALL individuals with a molecularly confirmed diagnosis of SMS can participate
  - This includes individuals with:
    - The common 17p11.2 deletion
    - An uncommon 17p deletion that includes *RAI1*
    - A pathogenic variant in *RAI1*
  - Genetic test results are required

# What is needed to participate in the patient registry?

- A copy of the genetic test results for the individual with SMS

Genetic test results can include:

- Karyotype
- FISH
- Chromosomal Microarray (CMA)
- *RAI1* Sequencing
- Gene Panel Testing (e.g., seizure or epilepsy gene panel or autism/intellectual disability panel)
- Rare Obesity Program Testing (Rhythm Pharmaceuticals)
- Whole Exome Sequencing
- Genome Sequencing

- Proof of parent/guardian identity (for minor children) or legal guardianship/ power of attorney (for adults)
- A “signed” consent form (now available online!)

# What is asked?

What is asked in the registry surveys?

What additional information can be collected in the registry?



# What is asked in the registry surveys?

- Questionnaires are completed online using a personalized link
  - Demographics
  - Medical history
  - Birth Measurements
  - Medication log
  - Body Type Assessment
  - Hyperphagia Questionnaire
  - Speech & Language Development
  - Food Related Problems Questionnaire
  - Behavior Problems Inventory
  - Child Sleep Habits

# What other information can be collected in the registry?

- Medically relevant information to promote better understanding of the natural history of SMS
- Examples include:
  - Sleep studies
  - Hearing tests
  - Spine x-rays
  - Photographs, videotapes, audiotapes
  - Questionnaires about sleep habits, quality of life, movement disorder, hunger, etc.

# Let's talk about the data

End-user's  
view of data  
collection

Data Storage

Data Access

# End-user's view of data collection

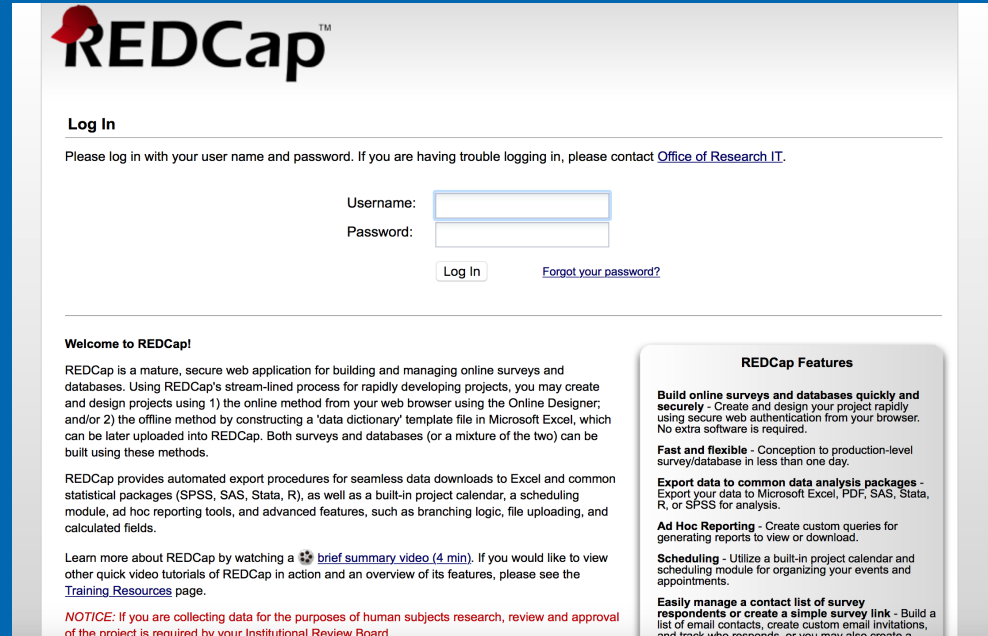
- You will receive an email containing a link to the first survey: Demographics
- The email will contain details about the option to “save & return later” or complete at one time
- Links to subsequent surveys will be emailed following the completion of the prior questionnaire
- How will this look?

[Demographics Survey](#)

[Returning to Demographics Survey](#)

# Where will my data be stored?

- All data will be saved on a REDCap server located at Baylor College of Medicine
- REDCap is a secure web application used for building and maintaining online surveys and databases



The screenshot shows the REDCap login interface. At the top left is the REDCap logo, which consists of a red pill icon followed by the text 'REDCap™'. Below the logo is a 'Log In' section with a heading 'Log In' and a sub-heading 'Please log in with your user name and password. If you are having trouble logging in, please contact [Office of Research IT](#).' There are two input fields: 'Username:' and 'Password:'. Below the 'Password:' field are two buttons: 'Log In' and 'Forgot your password?'. Below the login section is a 'Welcome to REDCap!' section with a paragraph of text describing the application. To the right of this section is a 'REDCap Features' box with a heading 'REDCap Features' and several bullet points describing features like 'Build online surveys and databases quickly and securely', 'Fast and flexible', 'Export data to common data analysis packages', 'Ad Hoc Reporting', 'Scheduling', and 'Easily manage a contact list of survey respondents or create a simple survey link'. At the bottom of the 'Welcome to REDCap!' section is a 'NOTICE' in red text: 'NOTICE: If you are collecting data for the purposes of human subjects research, review and approval of the project is required by your Institutional Review Board.'

**REDCap™**

### Log In

Please log in with your user name and password. If you are having trouble logging in, please contact [Office of Research IT](#).

Username:

Password:

[Log In](#) [Forgot your password?](#)

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**Welcome to REDCap!**

REDCap is a mature, secure web application for building and managing online surveys and databases. Using REDCap's stream-lined process for rapidly developing projects, you may create and design projects using 1) the online method from your web browser using the Online Designer; and/or 2) the offline method by constructing a 'data dictionary' template file in Microsoft Excel, which can be later uploaded into REDCap. Both surveys and databases (or a mixture of the two) can be built using these methods.

REDCap provides automated export procedures for seamless data downloads to Excel and common statistical packages (SPSS, SAS, Stata, R), as well as a built-in project calendar, a scheduling module, ad hoc reporting tools, and advanced features, such as branching logic, file uploading, and calculated fields.

Learn more about REDCap by watching a [brief summary video \(4 min\)](#). If you would like to view other quick video tutorials of REDCap in action and an overview of its features, please see the [Training Resources](#) page.

**NOTICE:** If you are collecting data for the purposes of human subjects research, review and approval of the project is required by your Institutional Review Board.

#### REDCap Features

- Build online surveys and databases quickly and securely** - Create and design your project rapidly using secure web authentication from your browser. No extra software is required.
- Fast and flexible** - Conception to production-level survey/database in less than one day.
- Export data to common data analysis packages** - Export your data to Microsoft Excel, PDF, SAS, Stata, R, or SPSS for analysis.
- Ad Hoc Reporting** - Create custom queries for generating reports to view or download.
- Scheduling** - Utilize a built-in project calendar and scheduling module for organizing your events and appointments.
- Easily manage a contact list of survey respondents or create a simple survey link** - Build a list of email contacts, create custom email invitations, and track who responds, or you may also create a

# Who will have access to my data?

- Currently only researchers at Baylor College of Medicine have data access
- Other researchers interested in SMS can have access to the data\* following an approved process
- The data will NOT be linked to your personal information
  - Personal information includes:
    - Name
    - Date-of-birth
    - Email address
    - Phone number

Ready to  
participate  
and add to  
the growing  
knowledge  
of SMS?

Start at the PRISMS  
website!

[prisms.org](https://prisms.org)

prisms

# QUESTIONS?

Send email:  
[prisms.registry@bcm.edu](mailto:prisms.registry@bcm.edu)





# Study to Better Characterize Food-Related Behaviors in SMS

- Drs. Elsea and Sisley are conducting focus groups this weekend to better understand food-related behaviors in the SMS population
- NIH funded study – National Institute of Child Health and Human Development
- Goal is to create a better questionnaire for clinical trial use
- Seeking caregivers of SMS individuals who are at least 6 years old
- Focus groups will be audio recorded for accurate transcription
  - Thursday at 3:30 in McMillan
  - Saturday at 12:30p in Beeman A
  - Additional virtual sessions after the conference
- For more information, contact Stephanie Sisley 513-465-8701 or email [Sisley.lab@bcm.edu](mailto:Sisley.lab@bcm.edu)