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2022

*Envisioning
the Possibilities*

Smith-Magenis Syndrome Patient Registry Update and Current Research Opportunities

Theresa A. Wilson, MS, RD

Lead, Clinical Research

Elsa Lab

Baylor College of Medicine

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SMS Patient Registry

- Happy 5th Birthday!
- Publications
- Current Enrollment
- Immediate Future Plans

Opportunities in Current Research

- Focus Group Discussion
- Biomarker Study

Happy 5th Birthday SMSPR!

First participant
joined in July 2017



May 2019: online
consent available



Most recent new
participant joined
in January 2022



Summer 2019:
added new surveys
& multiple
languages

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Published Data from SMSPR





ELSEVIER

Research in Developmental Disabilities

Volume 127, August 2022, 104257



Relationships between food-related behaviors, obesity, and medication use in individuals with Smith-Magenis syndrome

Anusha A. Gandhi ^a, Theresa A. Wilson ^a, Stephanie Sisley ^{b, c}, Sarah H. Elsea ^a  , Rebecca H. Foster ^{d, e}

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Current Enrollment

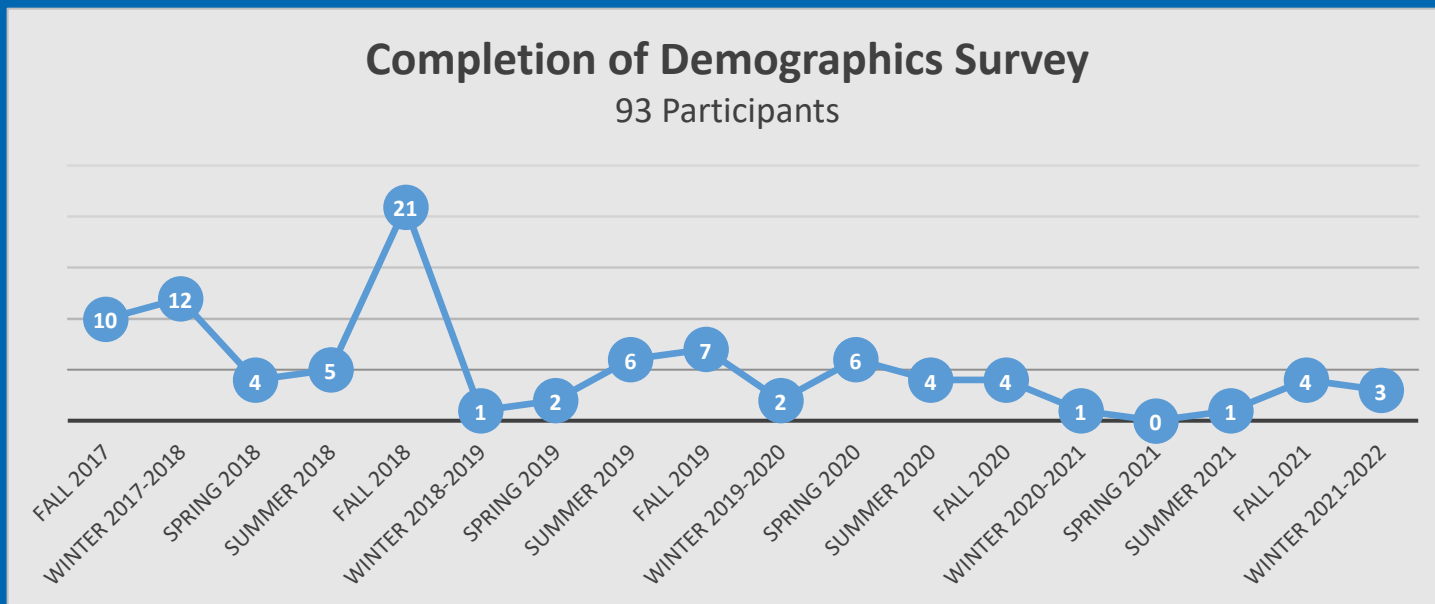
Enrollment
across the past
5 years

Where are our
participants?

Current snapshot of SMSPR participation

- Current enrollment is 102 families.
- Demographics complete for 93 families.
- We have had 231 inquiries about joining the patient registry, but only 46 completed.
- If you encounter a problem trying to sign up, PLEASE email prisms.registry@bcm.edu

SMSPR enrollment across time



Where are our participants?

https://app.mapline.com/map/map_60877551/PQoBYTAUHD8pHhZ5P3hwP2IUNj8IVGEUOjwUPz87AT9KPwgKP3

Immediate Future Plans

Convert database
to longitudinal
format

Thoughtfully add
useful surveys

Collaborations

Opportunities in Current Research Studies

Focus Group
discussion to
improve surveys
for SMS

Biomarker Study

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
- Study funded by NIH – 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
 - Saturday at 12:30p in Beeman A
 - Additional virtual sessions after the conference

For more information, contact Dr. Stephanie Sisley 513-465-8701 or email Sisley.lab@bcm.edu

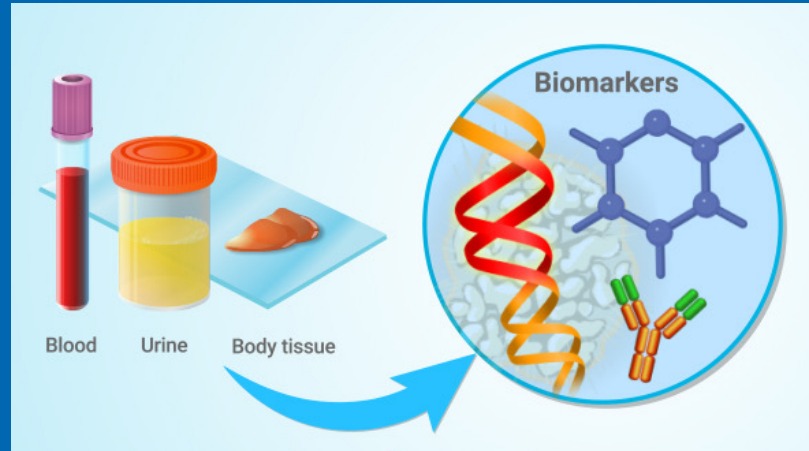


SMS Biomarker Study

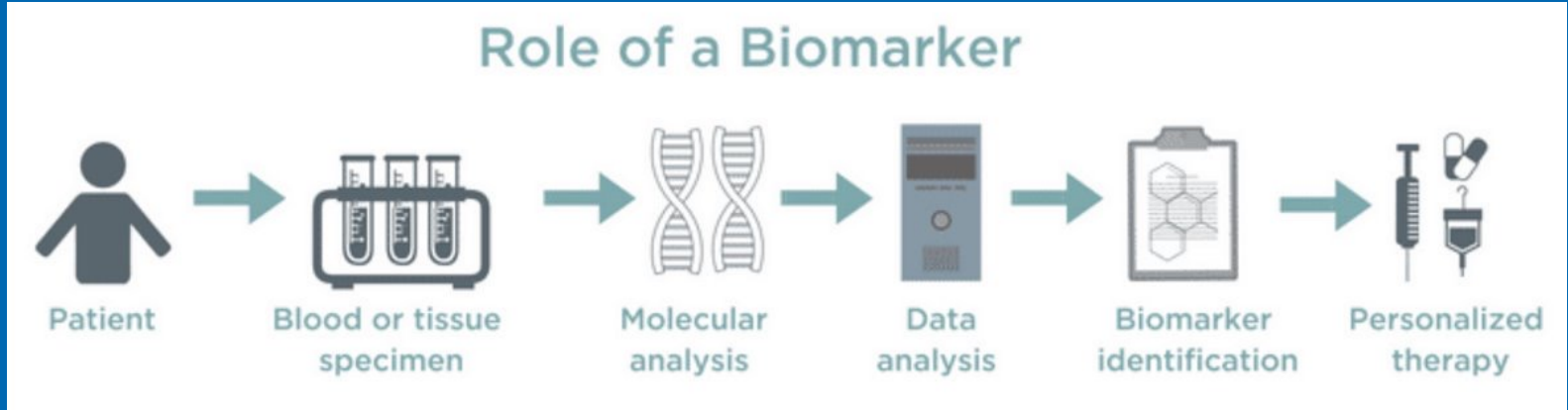
- Dr. Elsea is conducting a study to validate newly identified biomarkers in persons with SMS

What is a biomarker?

A biomarker can be a physiological, molecular, radiological or other finding that is associated with a specific condition.



SMS Biomarker Study



We are collecting blood and urine samples to investigate and validate potential new biomarkers that may help us better understand and monitor physiological differences in SMS.

SMS Biomarker Study

- Collecting blood and/or urine samples
- **Friday 1:00 – 3:00 in Rooms: Beeman A & McMillan**
- By pre-registration and appointment only
- Informed consent is required
- Please stop by our table before 1:00p in the **Exhibit Area** for more details



THANK YOU!

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