

Cover Letter

Official title: Developing a Down Syndrome Health Instrument

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Information Sheet for Study:

Developing a Down Syndrome Health Instrument

Principal Investigator: Stephanie Santoro, MD

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Although over 200,000 individuals with Down syndrome live in the United States, studies to date have focused on outcomes apart from health. We need to accurately measure the health of all individuals with DS – and there are not similar tools for this population available. The overall goal of this research study is to create an instrument that will provide a barometer of the current state of health for DS and hold use in future research. Through focus groups we have created an instrument that directly assesses health in DS – the Down syndrome Health Instrument (DHI). Now that the DHI has been created, it is our goal to measure its effectiveness.

This project has three parts:

1. **Get input:** To conduct focus groups among caregivers, individuals with DS, panels of experts on DS and primary care physicians, and cognitive interviews to refine a conceptual model of health for DS and create an item pool,
2. **Draft survey:** To administer the DHI and establish internal validity, reliability, and external validity of the DHI for use in clinical research, and
3. **Use the instrument:** To test the usability of the DHI in two pilot settings: research and clinical. This instrument will measure patient-reported health in DS for the first time and allow measurement of health as an outcome which is not currently possible in this population. This can identify gaps in care, then direct and optimize interventions that will improve care.

Measuring the effectiveness of the DHI is as equally important as its creation. Completing the DHI will allow us to analyze results and determine the validity of the instrument. We ask for your participation in this project by completing the survey. Our goal is to have 832 survey respondents. The DHI is a 70 question survey that can be completed electronically or on paper. As a thank you for completing the survey you will be given \$25. Results from the survey will be used for instrument validation purposes only. Participation is **voluntary** and can stop at any time prior to turning in the completed survey. Deciding not to participate won't affect medical care received at Partners now or in the future.

To be eligible, you must 1) be the primary caregiver of an individual with Down syndrome or autism spectrum disorder (and the individual must be <22 years of age and not have mosaic DS), 2) be ≥ 18 years of age, 3) be fluent in written and spoken English, and 4) be able to read and provide informed consent.

If you would like to learn more about this study, please contact Ashlee Campbell at (617) 643-3197.

- IRB contact information: "If you'd like to speak to someone not involved in this research about your rights as a research subject, or any concerns or complaints you may have about the research, contact the Partners Human Research Committee at (857) 282-1900."
- Sponsor of the research: none

We are required by the Health Insurance Portability and Accountability Act (HIPAA) to protect the privacy of health information obtained for research. This is an abbreviated notice, and does not describe all details of this requirement. During this study, identifiable information about you or your health will be collected and shared with the researchers

conducting the research. In general, under federal law, identifiable health information is private. However, there are exceptions to this rule. In some cases, others may see your identifiable health information for purposes of research oversight, quality control, public health and safety, or law enforcement. We share your health information only when we must, and we ask anyone who receives it from us to protect your privacy.

We appreciate your time to read about this study. Thank you in advance for your insight to help us learn more about health in individuals with Down syndrome!