

**Title:** DS-Connect: The Down Syndrome Registry, The Professional Portal and INCLUDE/Down Syndrome Research Plan

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**Background:** The National Institutes of Health (NIH) supports Down syndrome (DS) research to better understand the condition and develop treatments and interventions. NICHD/NIH launched DS-Connect®: The Down Syndrome Registry (<http://DSConnect.nih.gov>) in 2013 to facilitate information sharing among persons with DS, families, and researchers. It consists of a series of online surveys to collect demographic information as well as health information about people with DS. The Registry has about 5090 participants, and has been supported by the Down Syndrome Consortium, composed of self-advocates, family members, DS organizations and foundations, and the NIH. The registry will also facilitate linkages with other research studies using a Global Unique Identifier (GUID) which will allow us to build a large virtual DS cohort.

In 2014, the DS-Connect® professional portal was launched to allow approved investigators, clinicians or other qualified professionals interested in DS to access the de-identified data for research purposes. The registry allows approved researchers to use de-identified data to develop studies on the etiology, natural history, and treatments for DS and associated conditions under three levels of access. The DS-Connect® Research Review Committee (RRC) oversees access and use of the registry data for some level 2 and all level 3 purposes. Access level 1 is for investigators who want to view aggregate, de-identified data (what participants see, e.g., charts and graphs using basic filters such as gender and age ranges) or who want to review the survey questions and content. Level 2 access requires approval from the DS-Connect® coordinator (and in some cases, from the RRC). This level of access is for investigators who want to perform customized searches, propose new survey modules, request a letter of support, or view aggregate, de-identified data and receive .csv/excel reports of row-by-row de-identified participant data, or to perform statistical evaluation for analysis, publication or presentation. Level 3 access is for an investigator who wants to post a study announcement on the DS-Connect® website, recruit subjects for a research or clinical study through the DS-Connect® registry coordinator, or who wants to use the data for a commercial purpose.

DS-Connect® has supported about 55 research studies to use the registry for recruitment purposes. Some examples include:

1. Safety and efficacy of tofacitinib for immune skin conditions in Down syndrome
2. The Effect of Congenital Heart Disease on Employment, Neuropsychological Outcomes, and Quality of Life in Adults with Down Syndrome
3. Depression, Stress, and Down Syndrome. A Multimethod Approach to Assessment
4. Music in Children and Families
5. Identify medical needs among those living with Down syndrome
6. Determine study feasibility and plan for clinical trials and research studies
7. Assist with recruitment for studies and surveys
8. Use the registry to post study notices

In addition, NIH just collected many responses from the DS community to the Request For Information (RFI) to update the research plan for Down syndrome as well as The INCLUDE project. This plan would update the published 2014 research objectives for DS in Down Syndrome Directions: The NIH Research Plan on Down Syndrome. The DS/INCLUDE research plan will be made available on The INCLUDE Website soon.

**Methods:** The DS-Connect® registry has an online consenting process and all basic health and demographic information is collected using surveys. The website is accessible from mobile devices and is also available in Spanish. There are five easy steps to join DS-Connect®. This whole process takes about 15-20 minutes.

1. Go to <https://DSConnect.nih.gov>
2. Select the "Join the registry" link on the homepage.
3. An account is set up using a user name and a password. This will be used each time to log in to the DS-Connect account.
4. Then the online consent/assent form needs to be completed.
5. After filling in the basic demographic information about the person with DS, the Initial Health Questionnaire and other surveys needs to be completed.

**Conclusions:** DS-Connect® will help in meeting the goals of assembling a large study population of individuals with DS. A DS cohort is essential for following development of DS over time and fully characterizing the condition and its co-occurring conditions at different stages of development. This resource will also help to understand factors that may contribute to differential survival rates among different racial and ethnic groups. This goal is one of the key components of the new NIH

INCLUDE (INvestigation of Co-occurring conditions across the Lifespan to Understand Down syndromE) project, which was launched in June 2018 and aims to include people with DS in research that impacts them.

**References:** N/A