Considerations for clinical research participation

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Linda Crnic Institute for Down syndrome
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Our research mission:
To help people with Down syndrome live their happiest healthiest life
Each person is dealing with Down syndrome in his/her own unique, personal way.

They are more awesome than different!
Individuals with Down syndrome have alterations in disease incidence:

- Alzheimer’s disease
- Severe respiratory infections
- Leukemias
- Autoimmunity
- Autism
- Obstructive sleep apnea

The >400,000 Americans with Trisomy 21 may hold solutions to major medical conditions.
Trisomy 21, the molecular cause of Down syndrome, is defined by 3 copies of chromosome 21 rather than 2.
We know very little about how trisomy 21 causes Down syndrome.
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• Two types: observational studies and clinical trials.
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  – Clinical trials are research studies performed with people that are aimed at evaluating an intervention. Primary way that researchers find out if a new treatment is safe and effective in people.

https://www.nia.nih.gov/health/what-are-clinical-trials-and-studies
Anyone can participate in a wide variety of clinical studies:

- Treatment
- Blood draw
- Sleep study
- Speech intervention
Why do people participate in clinical studies?

• To help themselves/person with Down syndrome directly – either now or in the future.
  – May get new treatment for a disease before it is available to everyone else.
  – To play a more active role in your own health care.
  – Researchers may provide you with medical care and more frequent health check-ups as part of your treatment.

• To pay it forward and help people in the future.
  – You may help others get a better treatment for their health problems in the future.

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How to choose what clinical trials are right for you?

Balance

How to choose what clinical trials are right for you?

Cost & risk
- Side effects
- Psychological impacts
- Impacts on family

Potential benefit
- Quality of life
- More medical care
- Duration of life

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- **Phase II trial**: Uses more people (100 to 300). While the emphasis in Phase I is on safety, the emphasis in Phase II is on effectiveness. This phase aims to obtain preliminary data on whether the drug works in people who have a certain disease or condition. These trials also continue to study safety, including short-term side effects. This phase can last several years.

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- **Phase III trial**: Gathers more information about safety and effectiveness, studying different populations and different dosages, using the drug in combination with other drugs. The number of subjects usually ranges from several hundred to about 3,000 people. If the FDA agrees that the trial results are positive, it will approve the experimental drug or device.

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- **Phase IV trial**: For drugs or devices takes place after the FDA approves their use. A device or drug's effectiveness and safety are monitored in large, diverse populations. Sometimes, the side effects of a drug may not become clear until more people have taken it over a longer period of time.

How to choose what clinical trials are right for you?

Cost & risk
- Placebo
  - Psychological impacts
  - Impacts on family

Potential benefit
- Quality of life
  - More medical care
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Crnic Institute Human Trisome Project™

Minimally invasive observational study of the population with Down syndrome that employs cutting-edge multi-omics technologies for the analysis of various biological samples.

Cost & risk

Potential benefit

Balance

Crnic Institute Human Trisome Project™
(www.trisome.org)

Blood draw

Datasets

Medical Records & Participant Survey

Blood fractions

Digital Phenotypes

Proteomes, Exosomes

Metabolomes, Antibody Profiling

Genomes

Epigenomes

Transcriptomes

Proteomes

Functional Genomics

Functional Assays

Cell Line Generation

Immunophenotyping

Metabolomes

CBCs, blood smears

Differentiated Cell Types

Microbiomes

Urine Sample

Kidney Epithelial Progenitor Cells

Differentiated Cell Types

Monocytes

CD4+ T cells

CD8+ T cells

Natural Killer Cells

Granulocytes

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Natural Killer Cells

Granulocytes
The Power of Multidimensional Datasets

Going beyond the blueprint

Please visit poster #PO157 tonight for more details!
Crnic Institute Human Trisome Project™ (www.trisome.org)

Overall goals:

1. To define how trisomy 21 causes a novel disease spectrum.

2. To significantly accelerate research on Down syndrome.

3. To develop novel diagnostic and therapeutic tools that will benefit those with trisomy 21, and also millions of typical individuals.
Crnic Institute Human Trisome Project™ (www.trisome.org)

Short term goals:

1. To complete the most comprehensive cohort study of a population of individuals with trisomy 21 to date.

2. To create the largest public database for Down syndrome research to date.

3. To create the most comprehensive biobank of biological samples for Down syndrome research. To treat an existing ailment or disease.
Crnic Institute Human Trisome Project™ (www.trisome.org)

>520 participants enrolled

>320 individuals with Down syndrome

88 family units

39 states represented
Crnic Institute Human Trisome Project™ (www.trisome.org)
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Down Alzheimer Barcelona Neuroimaging Initiative (DABNI)

Dr. Juan Fortea Ormaechea

Laia Munoz Llahuna

100 samples of Cerebral spinal fluid
315 additional plasma samples of plasma
How to find relevant clinical studies

- Ask your physician or other local resources

- Websites
  - Clinicaltrials.gov
  - For example, we searched "Alzheimer's Disease, Down syndrome and United States" to find 9 relevant studies.
  - DSConnect.nih.gov
  - Connect with researchers and healthcare providers.
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Total of 7 clinical studies now recruiting on DS Connect

- 2 Studies of Aging and Alzheimer’s in Down syndrome: Alzheimer’s Clinical Trial Consortium (Dr. Michael Rafii). Clinical studies to identify biomarkers of aging and tracking progression of Alzheimer’s.

- University of Alabama Intellectual Disabilities Participant Registry: Regional registry to help participants find relevant research studies on language, learning, and memory that are low-risk to participants and not-for-profit.

- Brain Development in School-age Children with Down syndrome studied via brain MRI scan and behavioral/developmental assessments.

- Brain Development in Infants with Down Syndrome: MRI scan and developmental assessments.

- Patterns of Behavior in Developmental Disabilities in Genetic Conditions: online study is exploring the types of behaviors that children display throughout the day at home, at school, and in the community. One goal of the study is to create a new tool that can be used in future studies to better measure if treatments are effective for children with developmental and genetic conditions.

- The Relationship between Anxiety and Repetitive and Restrictive Behavior in Adolescents with Down Syndrome: studying the relationship between behaviors and mental health in children with Down syndrome.

- Stay tuned: LuMind RDS has recently launched a Down syndrome Clinical Trial Consortium Planning effort.
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“It’s a wild world…” even in basic research and clinical trials

• Consider the risks and benefits associated with any study
• The most important aspect is the well being of your loved one
• Thank you for your time