

Considerations for clinical research participation

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Linda Crnic Institute for Down syndrome
T21RS International Conference 2019: Barcelona
June 8th, 2019



Our research mission:

To help people with Down syndrome live their happiest healthiest life

Brother

Sister-in-law

Randy



**Each person is dealing with Down syndrome
in his/her own unique, personal way**



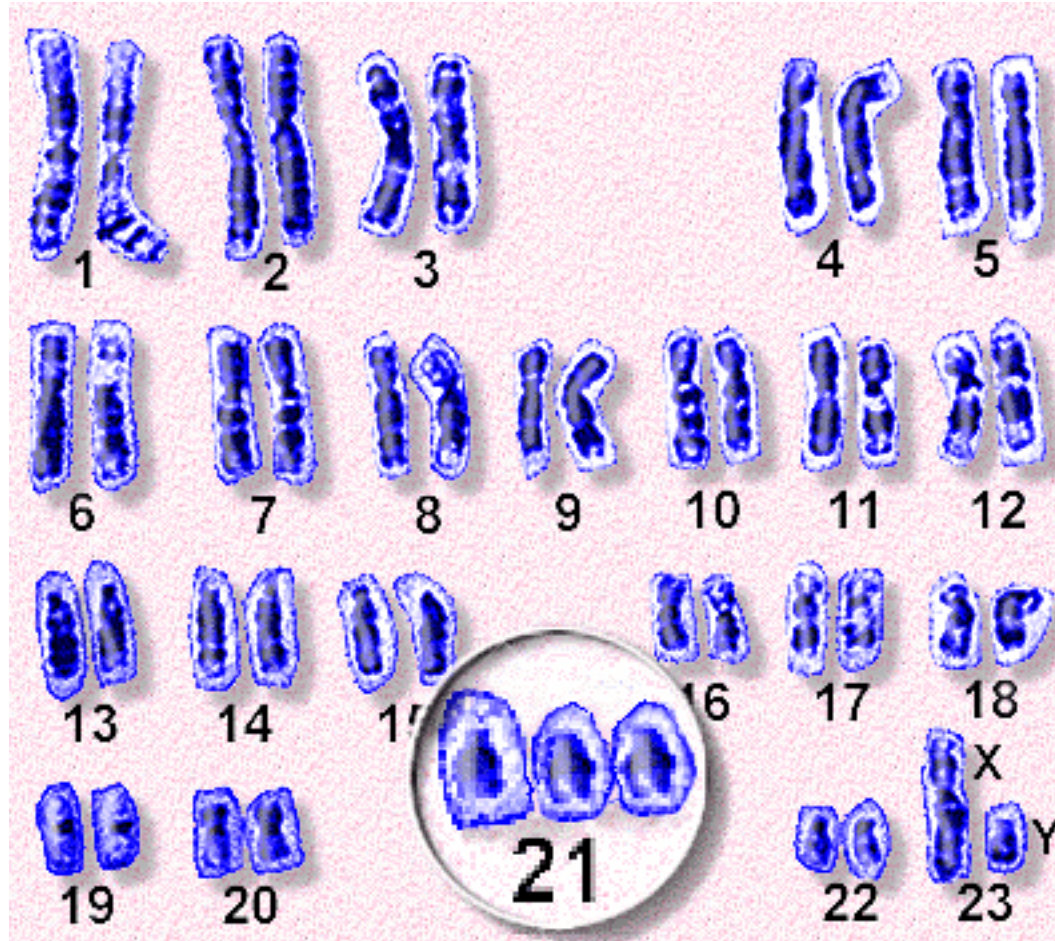
**They are more awesome than
different!**

Individuals with Down syndrome have alterations in disease incidence



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



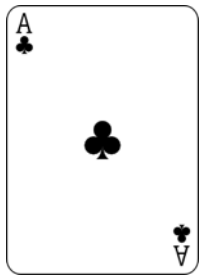
We know very little about how trisomy 21 causes Down syndrome

Trisomy 21



Down syndrome

Celiac Disease



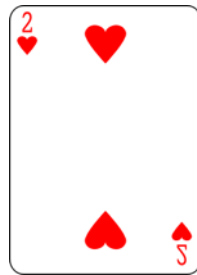
~5%

Intestinal Atresias



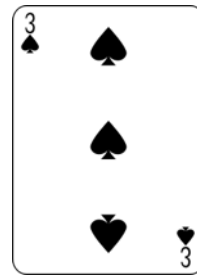
~12%

Thyroid Dysfunction



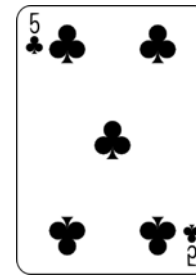
up to 18%

Seizures



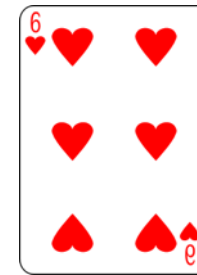
up to 13%

Leukemia



~1%

Congenital Heart Defects



40-50%

Autism



up to 10%

What are clinical trials and studies?

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- Two types: observational studies and clinical trials.
 - Observational studies observe people in normal settings without an intervention. These studies may help identify new possibilities for clinical trials.
 - 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.

Anyone can participate in a wide variety of clinical studies



Treatment



Blood draw



Sleep study



Speech intervention

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 - To treat an existing ailment or disease.
 - To prevent an ailment or disease.
 - 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.

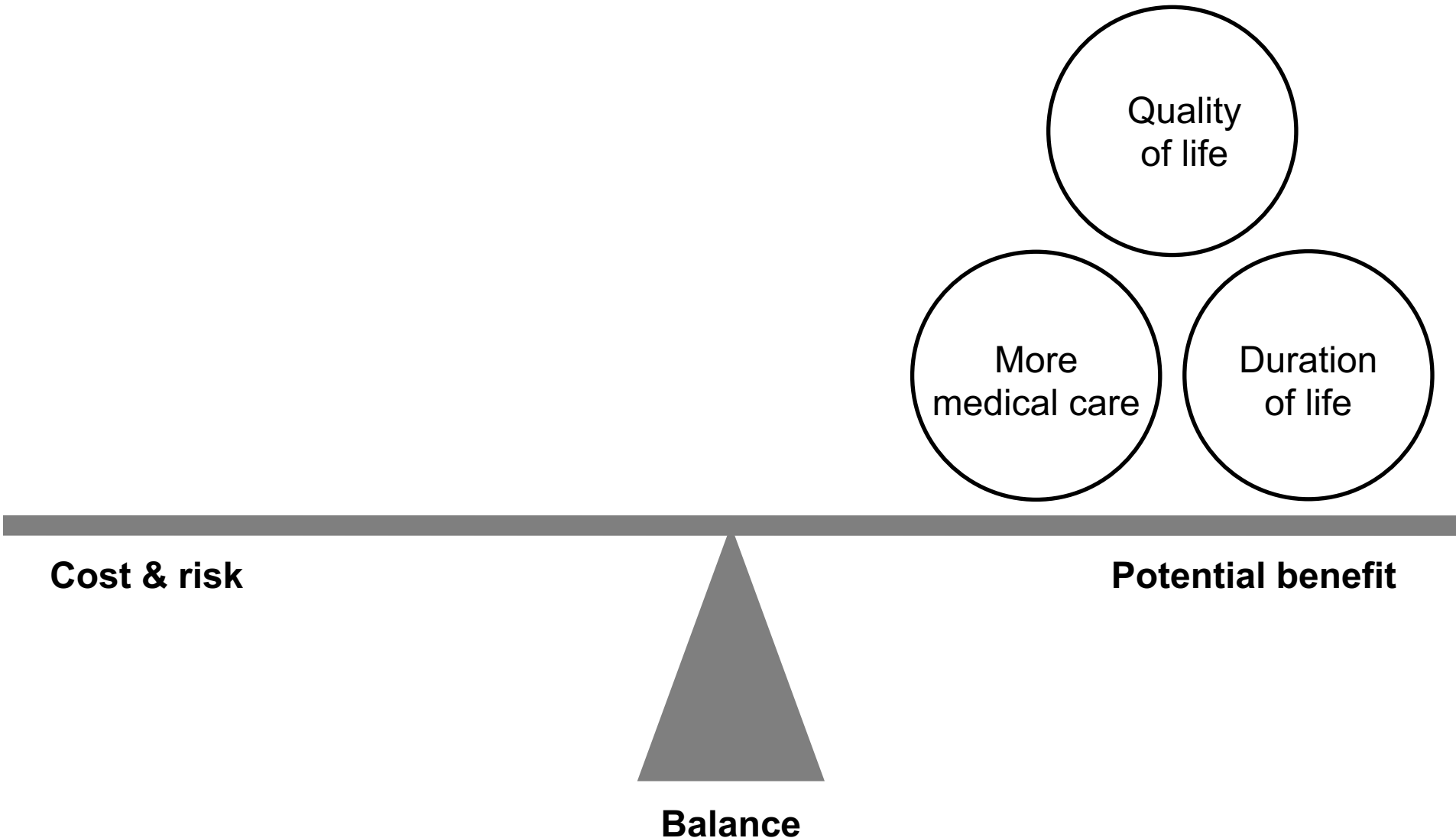
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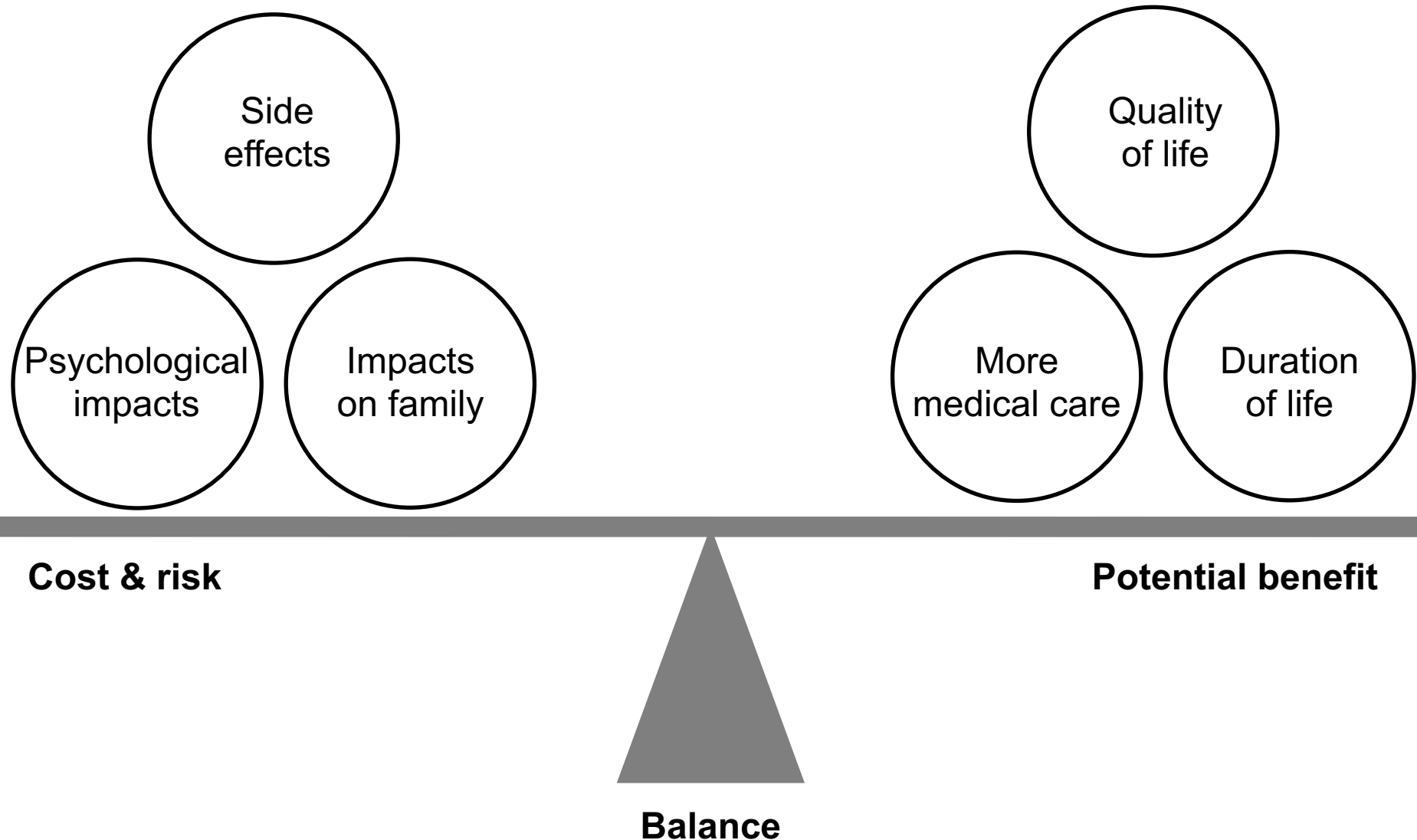
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- To pay it forward and help people in the future.
 - You may help others get a better treatment for their health problems in the future.

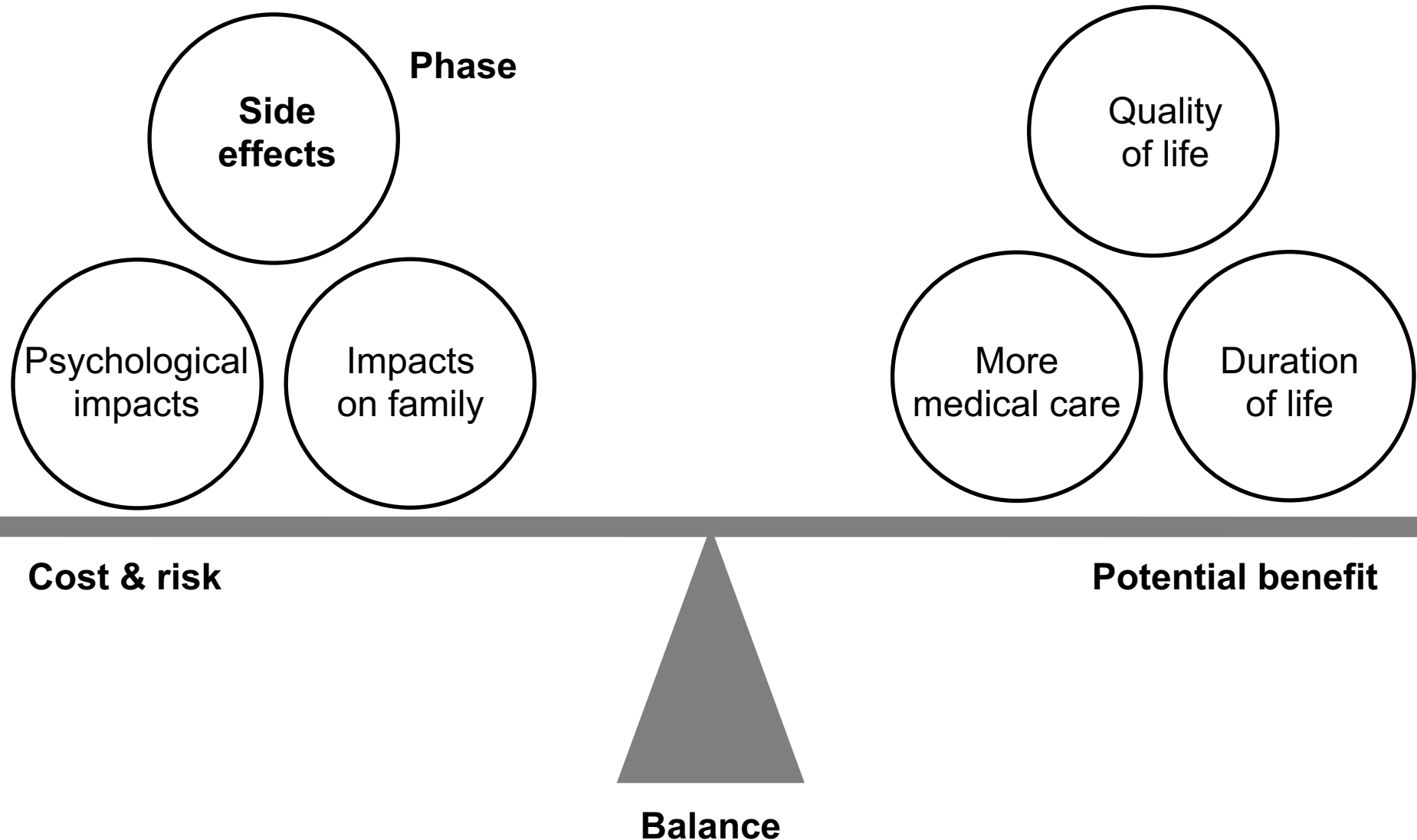
How to choose what clinical trials are right for you?



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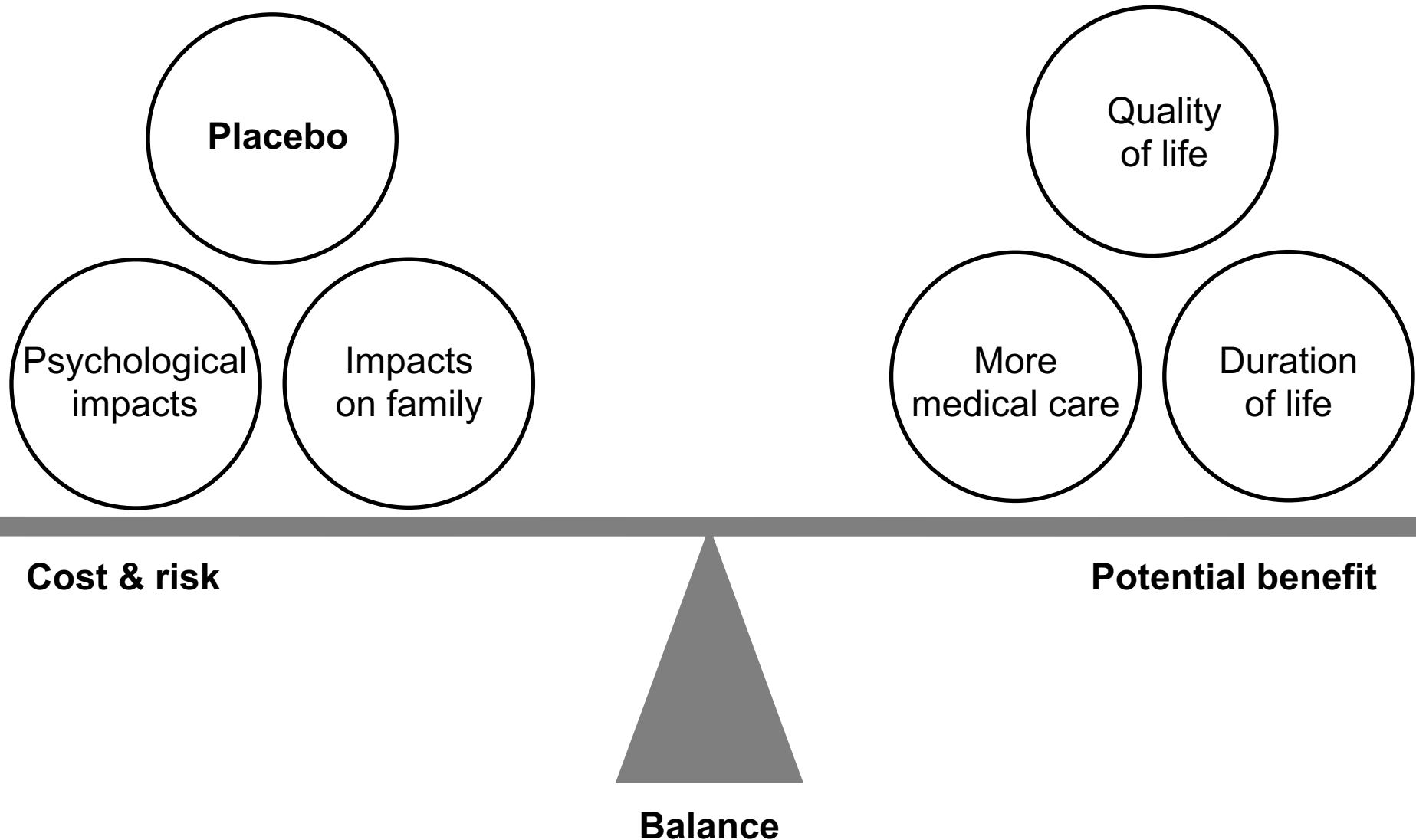
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- **Phase IV trial:** For drugs or devices takes place after the FDA approves their use. A device or drug's effectiveness and safety are monitored in large, diverse populations. Sometimes, the side effects of a drug may not become clear until more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

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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

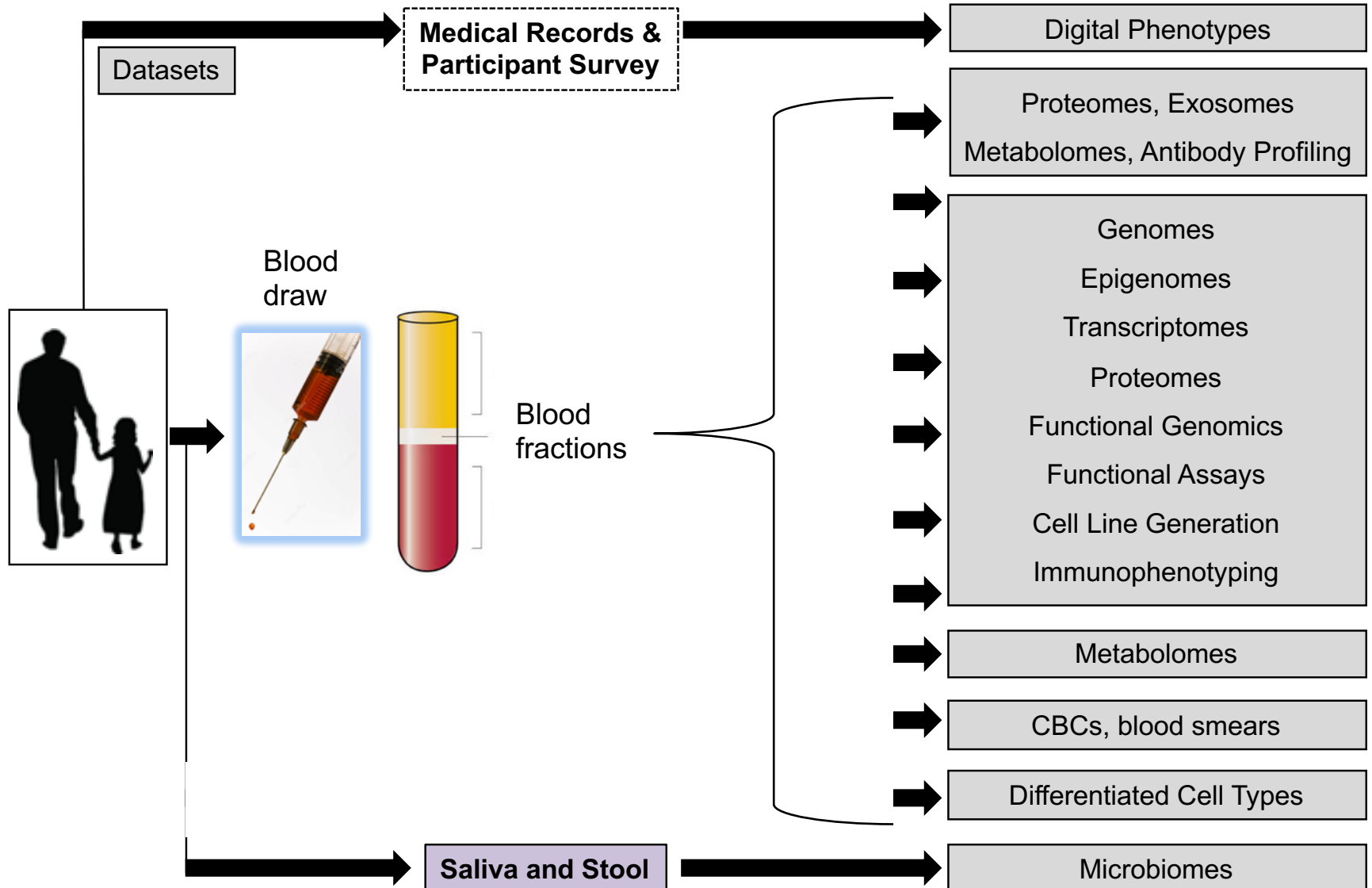
Potential benefit

Balance

<https://www.nia.nih.gov/health/clinical-trials-benefits-risks-and-safety>

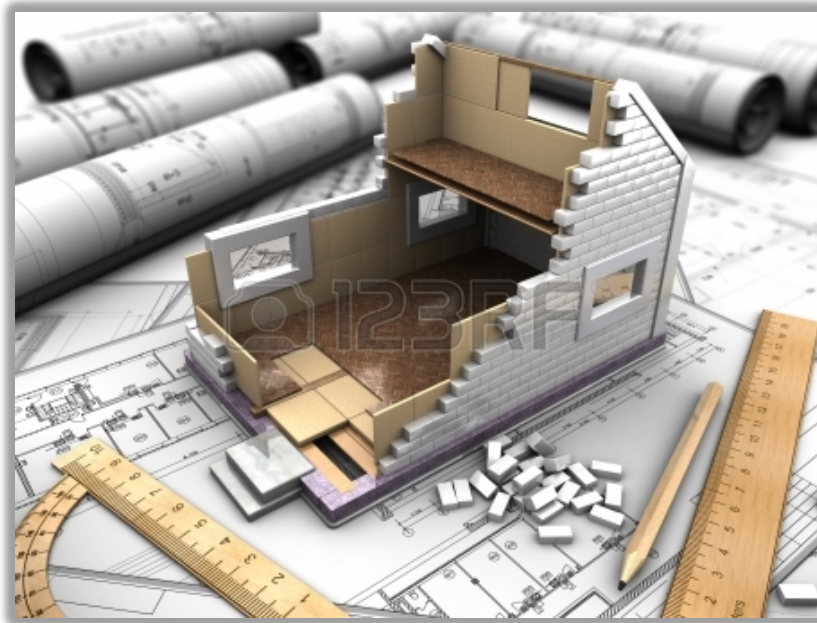
Crnic Institute Human Trisome Project™

(www.trisome.org)



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The Power of Multidimensional Datasets



Going beyond the blueprint

Please visit poster #PO157 tonight for more details!

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

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.

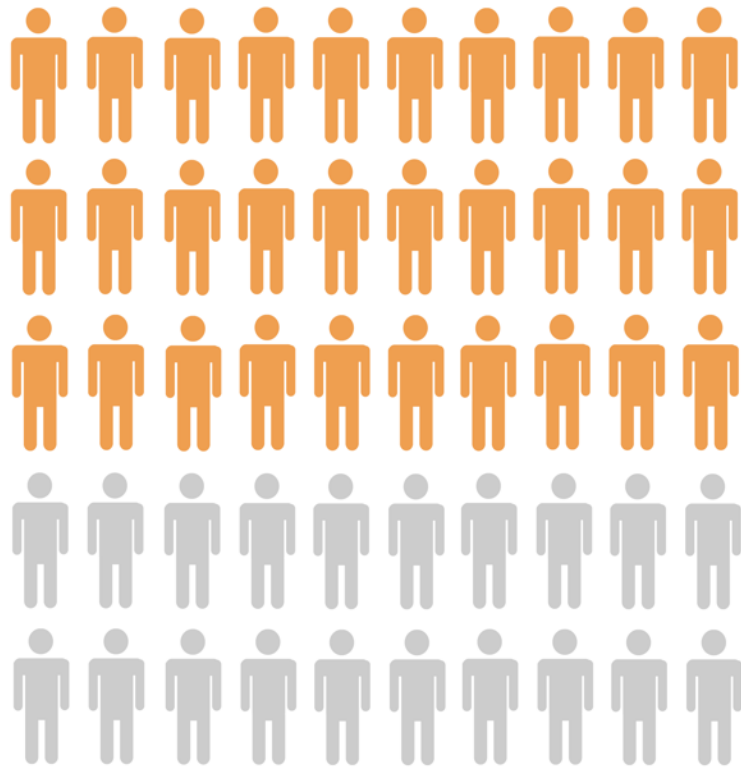
Crnic Institute Human Trisome Project™ (www.trisome.org)

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.

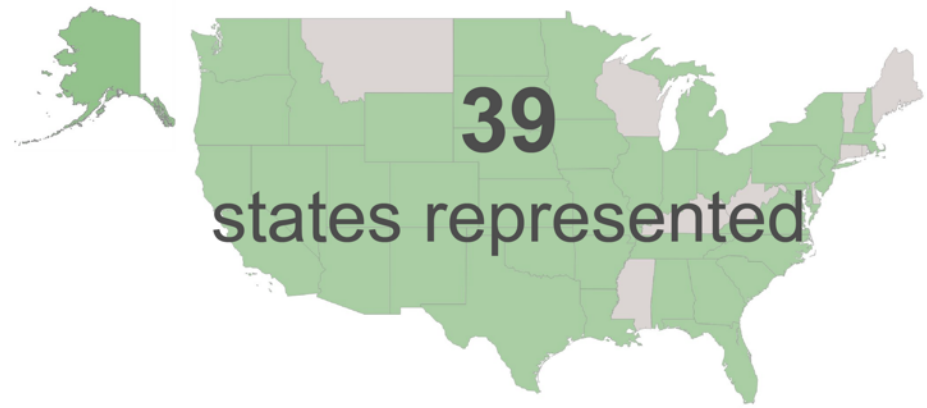
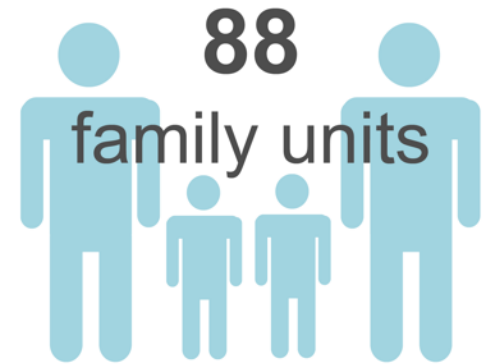
Crnic Institute Human Trisome Project™ (www.trisome.org)

>520 participants enrolled



>320

individuals with
Down syndrome



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

Barcelona 
Cataluna, Spain



maphill

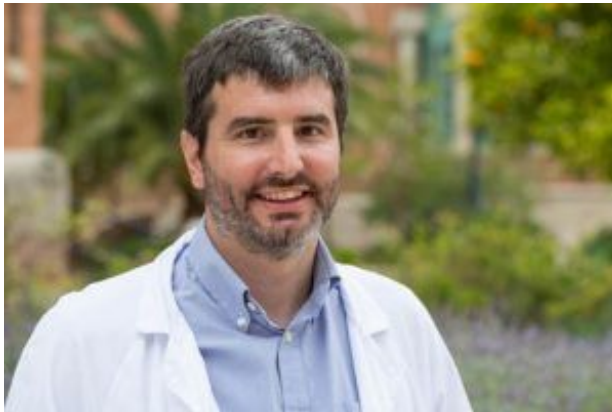


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Crnic Institute Human Trisome Project™ (www.trisome.org)

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

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- Websites
 - [Clinicaltrials.gov](https://clinicaltrials.gov).
 - For example, we searched “Alzheimer’s Disease, Down syndrome and United States” to find 9 relevant studies.
 - [DSCConnect.nih.gov](https://DSCconnect.nih.gov)
 - Connect with researchers and healthcare providers.
 - [DSCConnect.nih](https://DSCconnect.nih.gov).
- Ask scientists at this conference for international equivalents

Total of 7 clinical studies now recruiting on DS Connect

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- 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.

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- 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 Trials Network.*

“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

