



GLOBAL
DOWN SYNDROME
FOUNDATION®

The Importance for Science and Adult Medical Care to Work Together

Thursday, February 22, 2018

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Today's Presentation – We hope to answer some important questions...

❖ Global Down Syndrome Foundation's Adult Initiatives

- Medical Care Guidelines for Adults with DS
- Working together with community

❖ How Crnic Institute Research Can Support Medical Care

- Human Trisome Project

❖ The Importance of Medical Care for Research

❖ NEW Adult Down Syndrome Clinic at Denver Health

- Staffing and Services
- Research Priorities

❖ Q&A incorporating great advice we get at DSAIA



Adult Down Syndrome Initiatives

❖ Medical Care Guidelines

- The adult medical care guidelines were last updated with recommendations in 2001
- Since this time the lifespan and medical occurrences for people with Down syndrome have been updated and there is no way to get this crucial information to medical professionals



❖ Global is Committed To

- Working with medical and health professionals and with national health organizations
- Creating guidelines and a useful checklist “tool”
- Translating the guidelines and checklist tool into several languages
- Ensuring the guidelines are available free and easily to families, providers and Down syndrome organizations



The Importance of Adult Medical Care for Research

- ❖ **Through the process of creating the Guidelines, we will identify Gaps in our knowledgebase**
 - ❖ This is to be expected and important!
 - ❖ We don't know what we don't know
- ❖ **Once we identify the gaps, the Guideline working group and ECRI Institute will outline the study design necessary to address the gaps**
 - ❖ Global is committed to updating the guidelines every 5 years
 - ❖ The Guidelines will provide updated information and create a roadmap for future researchers to address knowledge gaps



How The Human Trisome Project Can Support Medical Care

- ❖ **Creating a biobank** of samples from people with Down syndrome
 - Will be able to find similarities and cohorts where you previously only had a single case study
- ❖ **Accessible for researchers and clinicians** who have limited or no access to people with Down syndrome
 - Evidence based data on the health of people with Down syndrome
- ❖ **Improving the lines of communication** between practitioners and researchers

**LINDA CRNIC
INSTITUTE
FOR DOWN SYNDROME™**

**RESEARCH TO DEVELOP THE
HUMAN TRISOME PROJECT BIOBANK**



PURPOSE OF THE STUDY
The purpose of this study is to provide qualified and approved researchers with access to biological samples and health information to answer specific research questions. This project will significantly increase the speed of Down syndrome research and the understanding of associated medical conditions such as leukemia and Alzheimer's disease.



WHO CAN PARTICIPATE?
Anyone who is 6 months – 89 years old who:
1. Has Down syndrome (any type, including mosaic or partial Down syndrome/trisomy21)
2. Has a family member with Down syndrome but does not him/herself have Down syndrome
3. Does not have Down syndrome and does not have a family member with Down syndrome (this would be a "control" group to compare with those affected by Down syndrome).



MAIN PROCEDURES INVOLVED
Participation would include a single successful blood draw, a mouth swab and allowing researchers to look at your health information. It is optional to give a stool sample and take part in the study for a longer time frame.

DURATION OF PARTICIPATION
A single visit to collect blood and a mouth swab, fill out a health survey, with the option of a stool sample and repeat visits for a longer term study.

INTERESTED?
Contact Angela Rachubinski at 303-724-7366
or hrp@ucdenver.edu
Compensation will be provided.
COMIRB#: 15-2170 PI: Joaquin Espinosa

Example of Breast Cancer in Adults with Down syndrome

Prevalence of breast cancer in DS compared to typical women

SIR = risk compared to typical women

Date	Authors	Study design	SIR	CI
2016	Hassle	Danish cytogenetics registry and Danish cancer registry 3,530 with DS. 3 cases “carcinomas at ages 31, 47, and 55 years”	0.16	0.03-0.47
2006	Patja	Finnish national registry of I/DD 3581 with DS. 6 cases	0.4	0.1-0.8
		Incidence for US women	12:100	5:100
		Incidence for US women with DS	2:100	

Example of Breast Cancer in Adults with Down syndrome

Prevalence

- May be 80% less common than in typical women

Treatment

- For some women with Down syndrome, any medical intervention is traumatizing

Test

- Many false positives
- Many of the true positives will detect minor tumors that would never cause harm

Pre-symptom stage

- Finding and treating a breast cancer late in a woman's life may not prolong her life

Risk to Women with Down Syndrome

Average Risk Women without Down syndrome			
	Number of mammograms/ averted breast cancer death	Number of mammograms/ LY gained	Number of false-positives/ averted breast cancer death
Biennial 50-74 (compared to no screening)	2,240	123	190
Annual 40-49, Biennial 50-74 (compared to Biennial 50-74)	7,896	235	953
Average Risk Women with Down syndrome			
Biennial 50-74 (compared to no screening)	16,735	2,752	1,493

More research needed!

- ❖ **Down syndrome changes the risk of developing many diseases**
- ❖ **Understanding why may help us understand these diseases better in everyone. We need to learn more!**

Less common

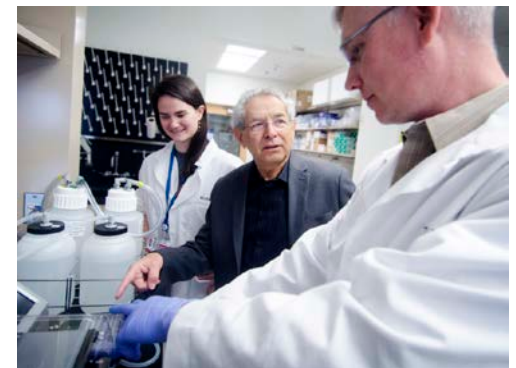
- ❖ **Cervical cancer:** are Pap tests helpful?
- ❖ **Colon cancer:** is screening helpful?
- ❖ **Heart attacks:** is screening for high cholesterol helpful?

More common

- ❖ **Thyroid disease:** is screening helpful?
- ❖ **Pneumonia:** should we have a more aggressive immunization schedule?
- ❖ **Alzheimer's disease:** is identifying early stages helpful?

Medical Care Guidelines for Adults with Down Syndrome

- ❖ **We need Global to provide leadership for the guidelines**
 - Strategy, logistics, legal support, translation interface, fundraising, and public relations associated with this important endeavor
- ❖ **We identified a national partner who has a roster of PhD/MDs and statisticians who can help us: ECRI Institute**
 - Professionally vet ideas versus recommendations versus guidelines
 - Get the guidelines published nationally
 - Has done this many times before
- ❖ **We have a team of MDs and other medical experts who specialize in ADULTS with Down syndrome and who can:**
 - Provide years of experience as an author or volunteer committee member
 - Who can commit 10% of their time for 2 years
 - Work as a team to ensure an excellent final product
 - Who are well-published in their fields and can help get an abstract of the guidelines published in high-impact journals
- ❖ **We need to identify expert medical translation companies to translate the final product into 10 languages**
- ❖ **Goal is to raise half the funds from the community and have Global support the other half!**
 - 36 Local Down Syndrome Organizations have already pledged support!
 - Plus more than 50 Individuals!



Adult Down Syndrome Clinic

❖ Why are we pushing for an adult clinic?

- Denver Adult Down Syndrome Clinic closed Feb 2017 and there is only 1 adult clinic in the 7 state region (Kansas)
- Pediatric Sie Center at CHC serves as pipeline for adult care
- Hub for basic and clinic research focused on Down syndrome in Colorado at Linda Crnic Institute
- Adult clinic would complete Global's affiliate model, and enhance our medical and research focus
- Adults with Down syndrome are underserved, misdiagnosed and face discrimination in medicine and research



Adult Down Syndrome Clinic at Denver Health

❖ Logistics:

- One day per week-Wednesday afternoons, 1:30pm-5:30pm
- Pena Primary Care Clinic
- 2 exam rooms
- Ages 21 and over with Down syndrome
- Continuity patients (30%) and consultative (70%)

❖ Services offered:

- Medical Care
- Behavioral Health
- Medical Social Work

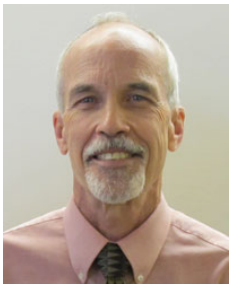


Adult Down Syndrome Clinic

at Denver Health: Staffing



Dr. Barry Martin, Lead Physician—Previous director of MCPN adult Ds clinic and current family physician employed by UCHHealth and privileged at DH



Dr. Dennis McGuire, LCSW, Behavioral Health—Completed credentialing and privileging process with Denver Health



Bryn Gelaro, LSW, Medical Social Worker—Completed HR and Badging process with Denver Health

Plus...

- ❖ Care Coordinator, TBD
- ❖ Clinic Navigator, TBD

Research Focus at the NEW Adult Clinic

- ❖ Denver Health's Commitment
- ❖ Global's Goals



How YOU Can Help

THANK YOU from the Global Family!

