

Sunday, July 7	
Time	
2:00-5:00pm	Check-In
5:00-6:30pm	Dinner (on your own)
6:00-7:00pm	New Family Orientation- Matt and Julie
7:00-8:30pm	Welcome Reception + Cash Bar
7:30-8:00pm	Performance by Bouncing Bulldogs Jump Rope Team

Monday, July 8				
Time	Track 1	Track 2	Self Advocates	Siblings
9:00-10:00am	Welcome Presentation w/ President Dave Aldrup, Neale Parker & Staff			
10:00-11:00am	Research Update with Dr. Cody	Ice Breakers & Transportation Presentation	Sibling Introductions (all ages)	
11:00-11:15am	Break			
11:15am-12:15pm	Dr. Karkar from the CRC - Neurology	Making Cards	Lynn's Hour (all ages)	
12:15-1:30pm	Lunch			
1:30-2:45pm	Special Local Guest Speaker - Molly Grantham "Anchor, Author, Speaker, Mom"			
2:45-3:15pm	Break			
3:15-4:15pm	Jenna Flynn- Autism Society	Dr. Catherine Larson- Child Psychiatry & C18 Sibling	Sue Hoss - Look, Cook, & Eat	Team Tie Dye
4:15-5:30pm	Syndrome Breakouts	Bingo!	TBD	
5:30-6:30pm	Free Time			
6:30-8:30pm	Meet in Lobby at 6:30 pm - Men's Night Out- Sweet Lew's BBQ/Music			
	Meet in Lobby at 3:30 pm - Siblings' Night Out - Carrowinds Amusement Park			
Silent Auction - Thursday morning to Friday 10:00 pm				

Tuesday, July 9				
Time	Track 1	Track 2	Self Advocates	Siblings
9:00-10:00am	Q&A w/ Dr. Dan Hale - Endocrinology	John McGing - SSI (tentative)	Crafts	Garrett's Hour w/ Donuts (+13)
10:00-11:00am	Ryan Platt- A Special Needs Plan	Hampton Hodges - A Special Needs Plan	Yoga	Parent Panel (+13)
11:00-11:15pm	Break			
11:15-12:15pm	Rick Guidotti - Positive Exposure		Painting	TBD
12:15-1:30pm	Lunch			
2:00-3:30pm	Supporting Chromosome 181 Dave Aldrup & Neale Parker	TBD	Crafts	Cyber Sib Call (all ages)
3:30-3:45pm	Break			
3:45-5:00pm	Men's Breakout Session	Women's Breakout Session	Scavenger Hunt & Karaoke	Embarrassment is Normal (+13)
5:00-6:30pm	Free Time			
6:30-8:30pm	Meet in Lobby at 6:30 pm - Ladies' Night Out-Sweet Lew's BBQ/Music			
	Meet in Lobby at 6:30 pm - SA Night - Polynesian Luau			

Wednesday, July 10				
Time	Track 1	Track 2	Self Advocates	Siblings
9:00-10:00am	Sibling Panel		SA Panel Prep	Mental Health Munchies/ Team Building (13+)
10:00-10:15am	Break			
10:15-11:45am	Self-Advocate Panel (Rick Guidotti moderator)			
11:45-12:00pm	Break			
12:00-1:30pm	Group Photos <span style="float: right;">Choose</span>			
1:30-4:00pm	Lunch on your own and Free Time (free admission to Carolina Raptor Center)			
3:00-4:00pm			Hair, Makeup and Princess Meet & Greet	Annual Scavenger Hunt
4:00-5:00PM	Free Time - Get Dressed & Ready for the Dance!			
5:15-6:30pm	Cocktail Reception, Silent Auction, and Meet & Great w/ The Top Cats cheerleaders from the Carolina Panthers			
6:30-10:00pm	Starfish Dinner & Dance			
Silent Auction checkout at 10:00pm (after Dance)				

## Enrollment Numbers

38418q-  
14018p-  
85 Tetrasomy 18p  
49 Ring 18  
37 Trisomy 18  
10 18q+  
5 18p+  
706Total

06/27/2023

## Enrollment Distribution

### North American Continent - 609

US (47 states) - 552

Canada (8 provinces) - 49

Mexico - 5

Barbados - 1

Puerto Rico - 1

Dominican Republic - 1

### South American Continent - 7

Brazil - 6

Columbia - 1

### Europe (20 countries) - 57

### Asia (4 countries) - 6

Australia - 22

New Zealand - 7

Africa (1 country) - 2

06/27/2023

# Chromosome 18

263 Genes



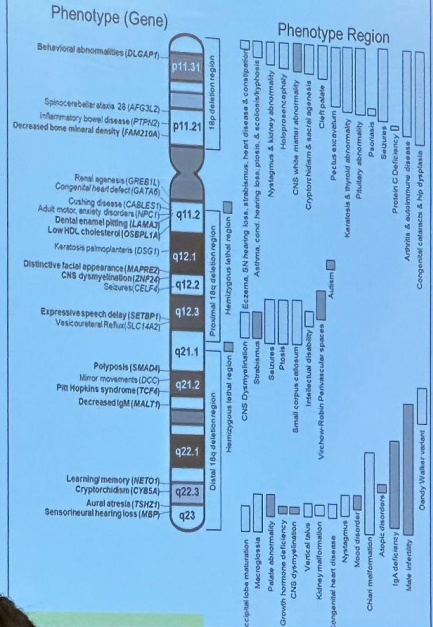
18p- &  
Tetrasomy 18p  
region  
67 genes

Proximal 18q-region  
87 genes

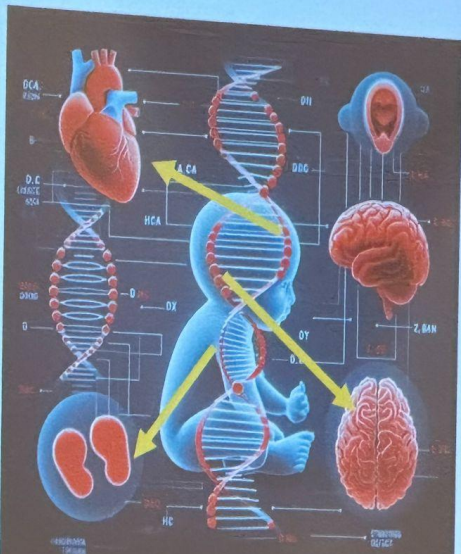
Distal 18q-region  
102 genes

# Molecular Genetics of the Chromosome 18 deletions

State of the Science 2022

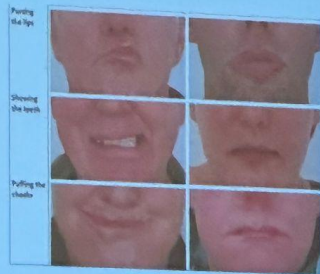


# GMKF - Linking Genes to Structural Defects



# Video Assessments of FSHD2

## FSHD2 Smile & Frown



## Facial Gesture Tests



Drs. Karkar & Tawil

Loonen, T.G.J., Horlings, C.G.C., Vincenot, S.C.C. et al. Characterizing the face in facioscapulohumeral muscular dystrophy. *J Neurol* 268, 1342–1350 (2021).

## Adult Onset Conditions

- 18p-

- FSHD2 } Gene

- SCA28 }

- Dystonia } Gene/ Symptom

- Arthritis } Symptom

- 18q-

- Hypothyroid } Symptom

- Autoimmune }



# All Participants

- Follow-up seizure survey 10 years
  - Dr. Karkar
- Annual surveys including medications
- Adult assessments
  - Living Arrangements
  - Educational Attainment
  - Work Status

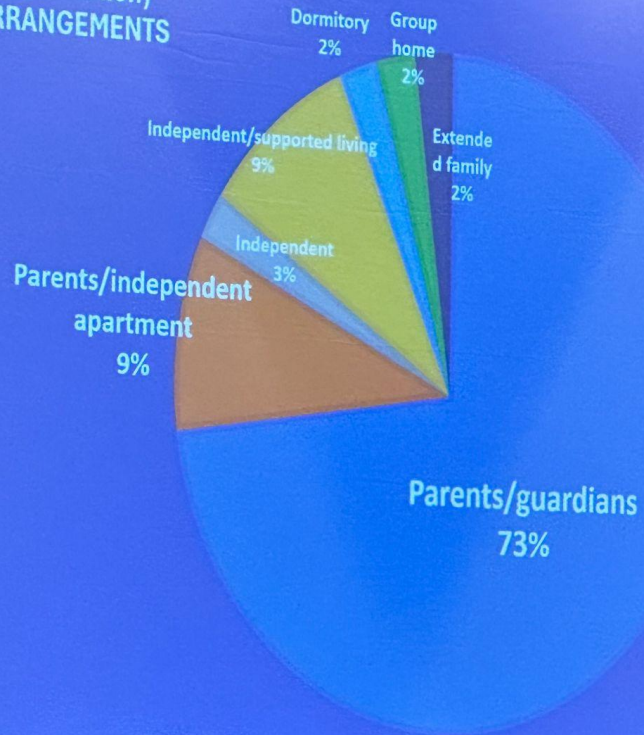
## 18p- (p arm deletion)

- Total no. of living participants over 18: N=91
- Total no. of participants we contacted: N=83
- Total no. of received Responses: N=44
- Deceased: N=4

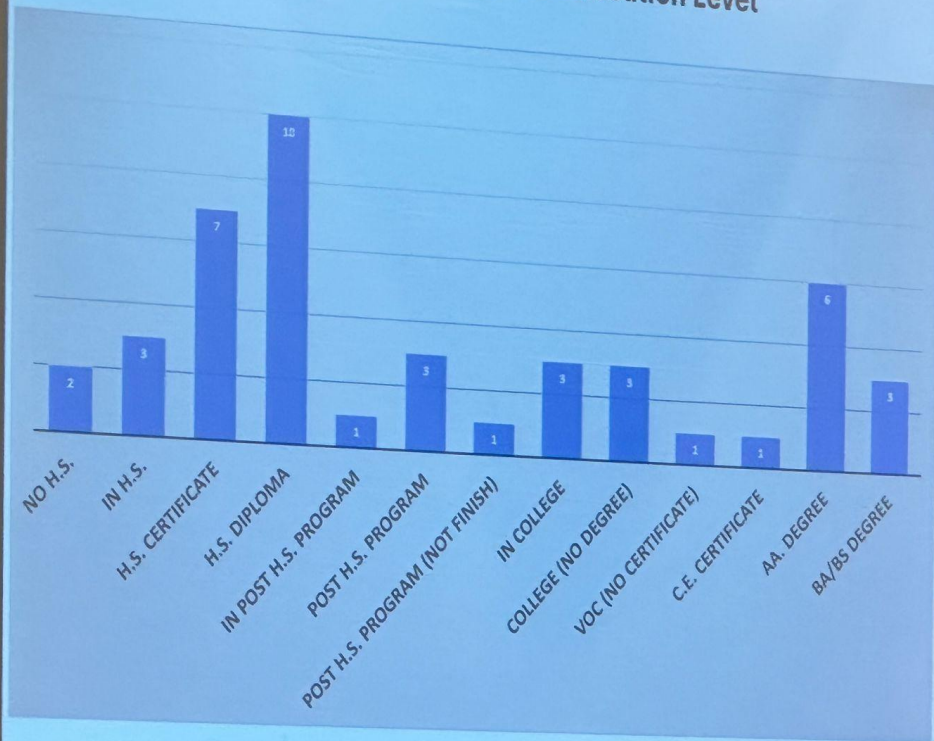
MARRIED (YES)	1
MARRIED (NEVER)	43

CHILDREN (YES)	0
CHILDREN (NO)	44

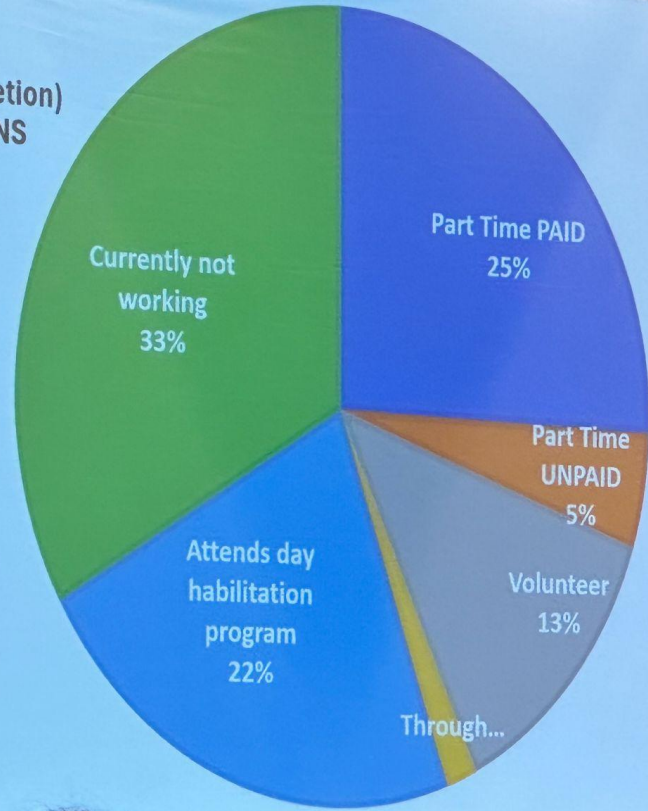
18p- (p arm deletion)  
LIVING ARRANGEMENTS



## 18p- Deletions Highest Education Level



18p- (p arm deletion)  
WORK POSITIONS



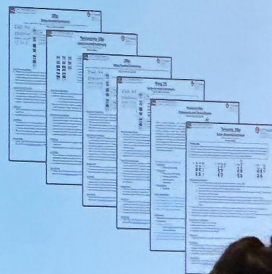
## Resources

### Registry Website

- Revised condition descriptions
- Introductory videos

### Chr 18 Clinical Research Center Website

- **Management Guides**
  - 18p-
  - Proximal 18q-
  - Distal 18q-
  - Ring 18
  - Tetrasomy 18p
  - Trisomy 18p
- **Gene Dosage Maps**



### Monthly videos and articles

## At this conference

- Blood draws for enrollment

Enrolled participants > 12 years old

- Smell test: 18q- & 18p-
- Video for speech and FSHD2: 18p-































