

# Frontiers in Congenital Disorders of Glycosylation – Natural History Project

July 2019

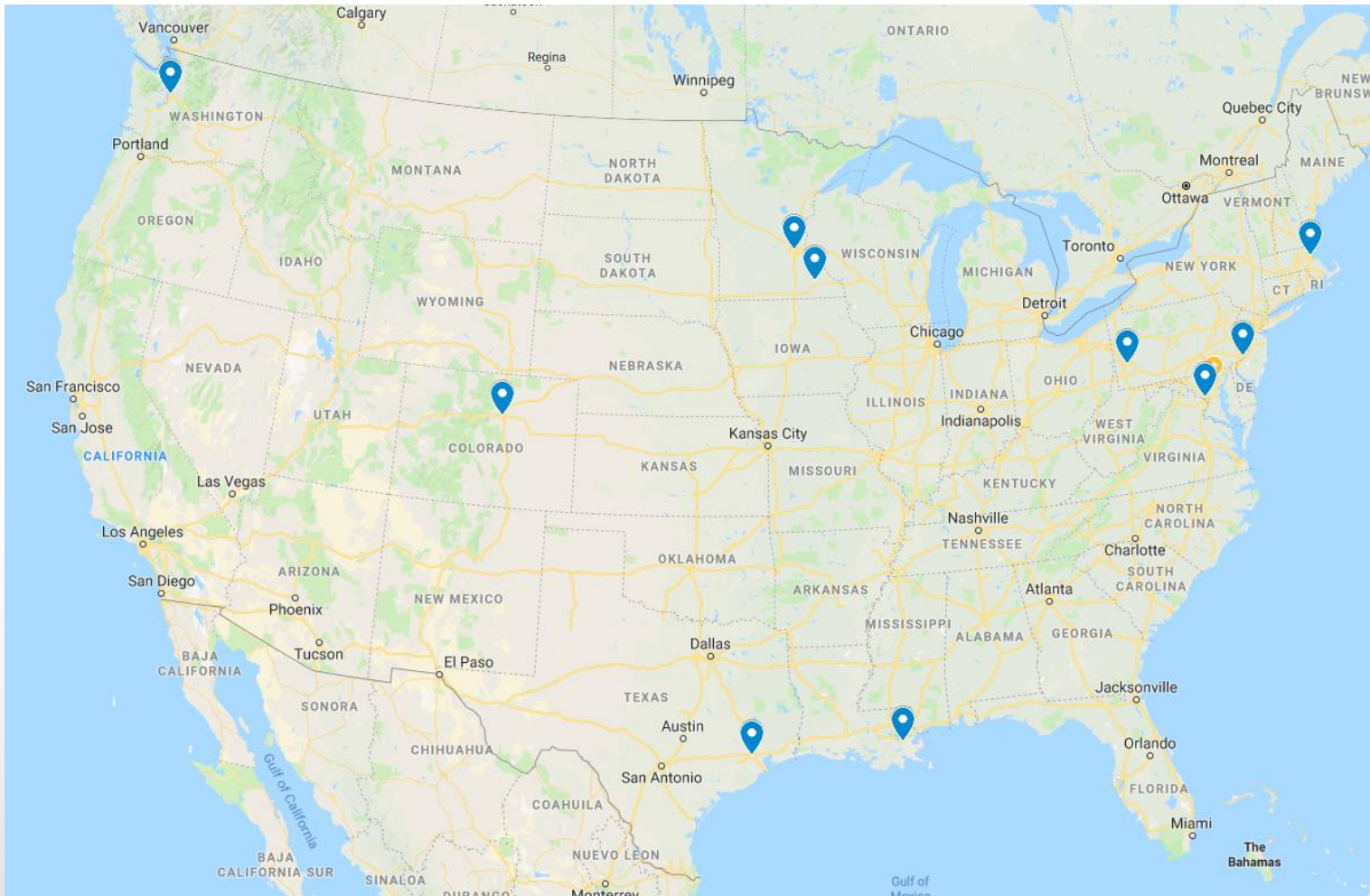
Lisbon, Portugal

# Specific Aims

- 1. Establish the prevalence and severity of specific morbid indicators of disease severity such as specific organ system involvement, degree of cognitive disability, and case-fatality associated with various CDG, and establish a dynamic platform to effectively disperse clinically relevant findings to families, non-expert clinicians and researchers, as well as provide a verified method to link these individuals to experts in CDG**
- 2. To validate the CDG rating scale and patient reported outcome /quality of life measures for CDG**

# Methods – Sites

1. Seattle Children's Hospital, Seattle WA
2. University of Colorado, Denver CO
3. Baylor College of Medicine, Houston TX
4. University of Minnesota, Minneapolis MN
5. Mayo Clinic, Rochester MN
6. Tulane University, New Orleans LA
7. National Institutes of Health, Bethesda MD
8. Children's Hospital of Pittsburg, Pittsburg PA
9. Children's Hospital of Philadelphia, PA
10. Boston Children's Hospital, Boston MA



# Methods

- Recruit All CDG Types
- Baseline and annual follow up visits; Travel reimbursed to a limit
  - Medical records collection
  - Interview with the family and patient
    - Detailed medical history
    - Nijmegen Pediatric CDG Rating Scale
    - Goal Attainment Scaling Scoring
  - Physical examination
  - Blood tests
  - Urine tests
  - Neurocognitive testing – Vineland Adaptive Behavior Scales, 3<sup>rd</sup> Ed
  - Clinical photography
  - \*Quality of life questionnaires
  - \*\*Skin Biopsy
- Design information sharing website and partner with CDG-Care PIN
- Will make all participants aware of CDG-Care PIN

\* - additional info available ; \*\* - optional, separate consent needed, only performed once

# Patient Reported Outcomes

- PROMIS questionnaires
  - Parent Proxy Bank v2.0 – Fatigue (23 Q), Parent Proxy Bank v2.0 – Mobility (23 Q) , Parent Proxy Bank v2.0 - Upper Extremity (29 Q), Parent Proxy 49 Profile (49 Q), Parent Proxy Scale v1.0 – Global Health 7+2 (9 Q), Parent Proxy Bank v1.0 - Physical Activity (10 Q), Parent-Proxy Bank v1.0 - Physical Stress Experience
- CDG Quality of Life Questionnaire being developed by CDG & Allies – PPAIN
- CDG Connect Patient Insights Network (PIN) questionnaires
  - 1) a Diagnosis survey, 2) a Family History questionnaire, 3) a General Health survey and 4) a Comprehensive Medical Care Questionnaire
- Goal Attainment Scaling

Parent Proxy Global Health 7+2

Please respond to each question or statement by marking one box per row.

		Excellent	Very Good	Good	Fair	Poor
Global01_PXR1	In general, would you say your child's health is:.....	<input type="checkbox"/> 5	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1
Global02_PXR1	In general, would you say your child's quality of life is:.....	<input type="checkbox"/> 5	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1
Global03_PXR1	In general, how would you rate your child's physical health? .....	<input type="checkbox"/> 5	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1
Global04_PXR1	In general, how would you rate your child's mental health, including mood and ability to think? .....	<input type="checkbox"/> 5	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1
		Never	Rarely	Sometimes	Often	Always
PedGlobal2_PXR1	How often does your child feel really sad? .....	<input type="checkbox"/> 5	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1
		Always	Often	Sometimes	Rarely	Never
PedGlobal3_PXR1	How often does your child have fun with friends? .....	<input type="checkbox"/> 5	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1
PedGlobal5_PXR1	How often does your child feel that you listen to his or her ideas? .....	<input type="checkbox"/> 5	<input type="checkbox"/> 4	<input type="checkbox"/> 3	<input type="checkbox"/> 2	<input type="checkbox"/> 1
	<b>In the past 7 days...</b>					
		Never	Almost Never	Sometimes	Often	Almost Always
PHfatigue2r	My child got tired easily .....	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4	<input type="checkbox"/> 5
PGpain5r	My child had trouble sleeping when he/she had pain.....	<input type="checkbox"/> 1	<input type="checkbox"/> 2	<input type="checkbox"/> 3	<input type="checkbox"/> 4	<input type="checkbox"/> 5

Example  
PROMIS  
Questionnaire

# How is this different from the Intramural NIH natural history (NH) study and the Glycomine NH study?

## Previous NIH NH Study

- Single Site
- Many more procedures and evaluations
- Each evaluation is longer (~ 1 week long)
- Data collected by NHGRI NIH

## Glycomine NH Study

- Different sites with some overlap
- Only studies PMM2-CDG
- Fewer procedures and evaluations at each evaluation
- Data collected every 6 months
- Data collected by Glycomine, Inc.

# Can you participate in this study as well as the other 2 studies?

- YES
- At this point, participating in one study does not preclude participation in the other studies (ie it is “Ale Carte”)
- For this study, participating in the other two studies are complimentary
  - As long as you sign a release of medical information to share test/evaluation results from the other studies/clinical evaluations, those tests/results will be collected and do not need to be repeated as long as they were done within 2 months of the study visit
  - Many sites participate in more than one of these studies



# Data will be entered into a Database/Registry

- Details are still pending and being developed
- Usage of iCloud to store data?
- Data sharing and ownership issues
- Ability to anonymously share data with whom?
  - Possibility of recontact
- Direct interface with families

Questions??