

## SSIEM Official Satellite Symposia

**“The 2nd World Conference on Congenital Disorders of Glycosylation (WCCDG)  
for Families and Professionals: a challenging story of sugar trees”,  
28 to 30 August 2015, in Lyon (France)**



**CONGENITAL  
DISORDERS OF  
GLYCOSYLATION  
WORLD CONFERENCE**

The power of advancing patient-oriented research united  
FAMILIES AND PROFESSIONALS

**This conference is part of the Educational Program of Excellence on CDG created by the Portuguese Association for CDG (APCDG, [www.apcdg.com](http://www.apcdg.com)).**

**It is organized in partnership with several associations and/or country CDG patient advocates: CDG Australia, CDG Brazil, CDG Czech Republic, CDG Denmark, Foundation of Glycosylation (the FoG) Canada, CDG Denmark, CDG Italy/Ireland, CDG Israel, Les ptits CDG France, CDG Spain, CDG Sweden, CDG USA, CDG UK charity and CDG The Netherlands.**



# How A Genetic Counselor Can Help CDG Families: Lessons from the Undiagnosed Diseases Program

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# Disclosures

I have no financial conflicts of interest to disclose.

The opinions I express in this presentation are personal and do not reflect an official endorsement by the U.S. Government, NIH, or NHGRI.



# What is Genetic Counseling?

“Genetic counseling is the process of helping people understand and adapt to the medical, psychological and familial implications of genetic contributions to disease”

*-National Society of Genetic Counselors*

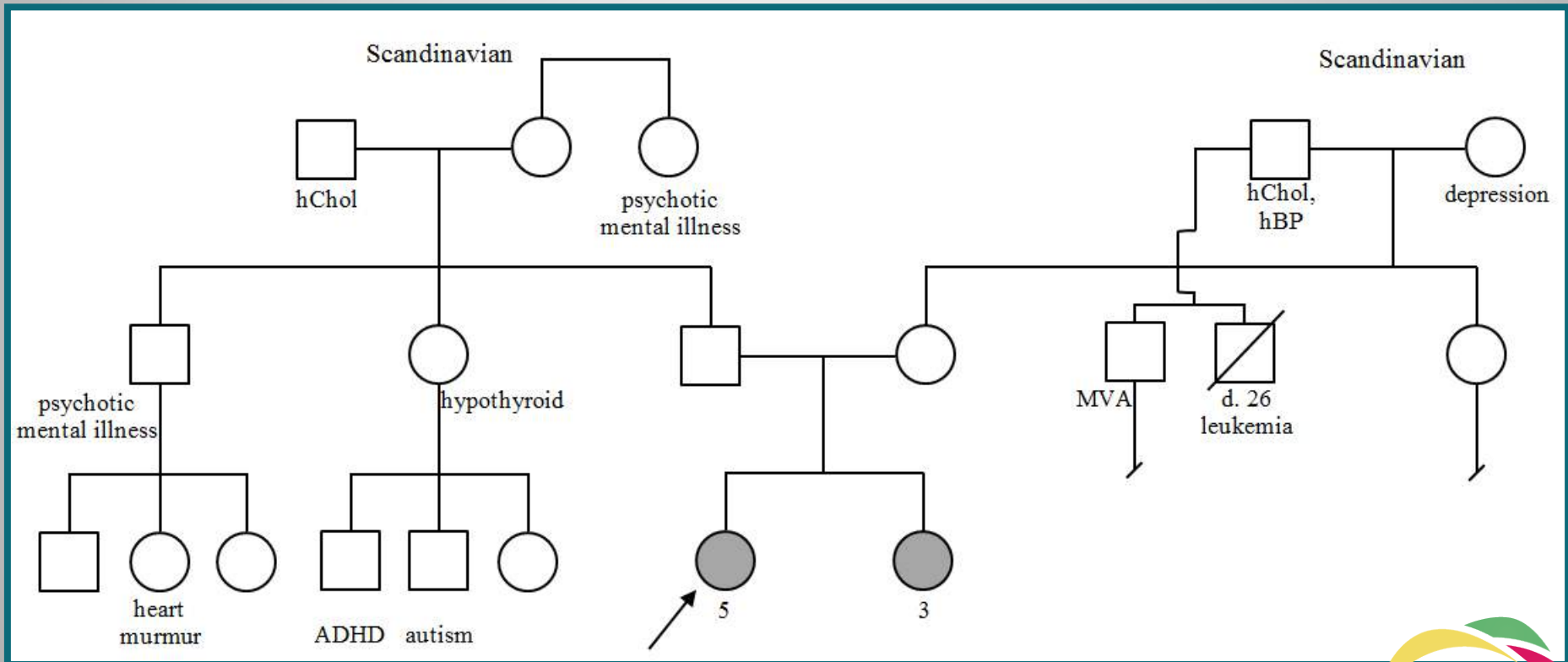


# What is a Genetic Counselor?

- **Have** a genetic condition
  - **At risk** of having a genetic condition
  - **Suspected** to have a genetic condition
- Facilitate understanding of the condition and what it means to have a genetic condition



# Case Example from the UDP



# Clinical Description

## 5 year old

- Developmental delay
- Low muscle tone
- Severe hypoglycemia
- Sleep disorder
- IgE elevation
- Intracranial hypertension

## 3 year old

- Developmental delay
- Low muscle tone
- Sensorineural hearing loss
- Severe hypoglycemia
- Central apnea
- Dysautonomia





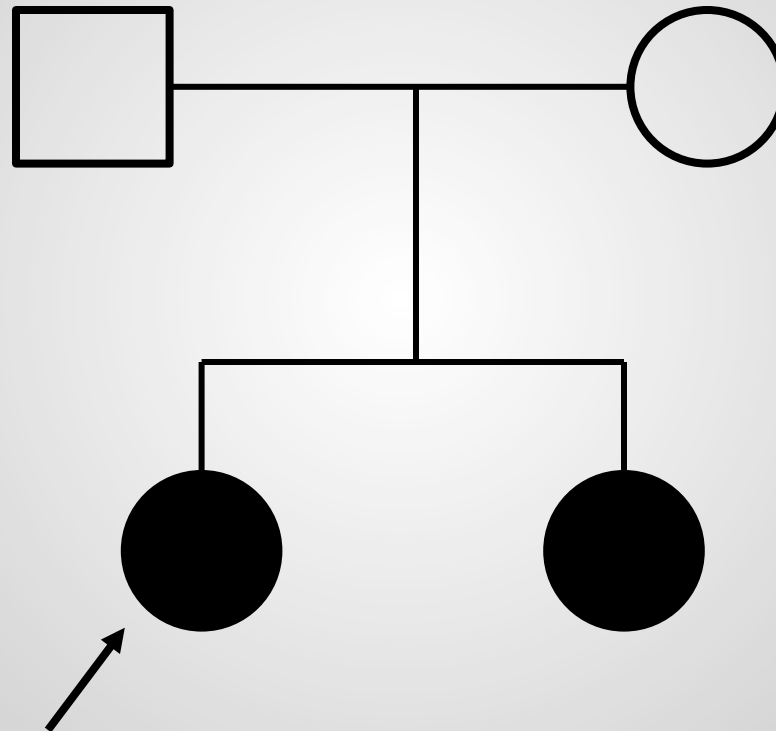
# Differential

- Disorders of Glycogen Storage or Carbohydrate Metabolism
- Disorders of Hormone Regulation
- Mitochondrial Diseases
- Congenital Disorders of Glycosylation

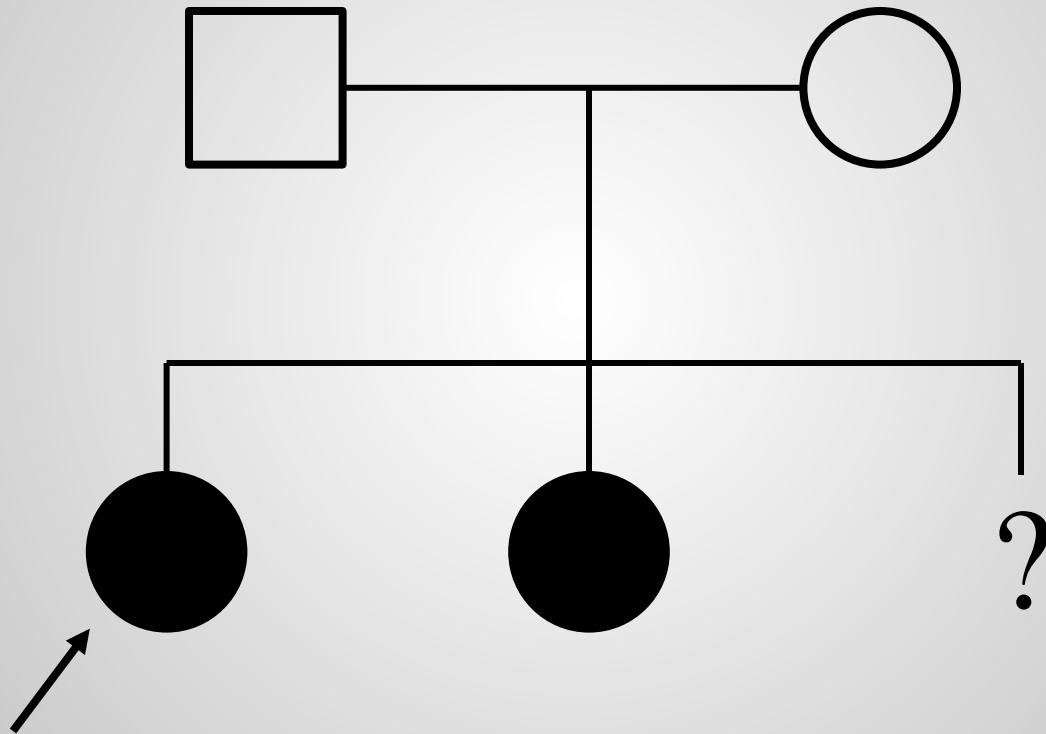
To date, extensive work-ups, including muscle & liver biopsies, whole exome sequencing, and mitochondrial genome testing on both girls have been unrevealing.



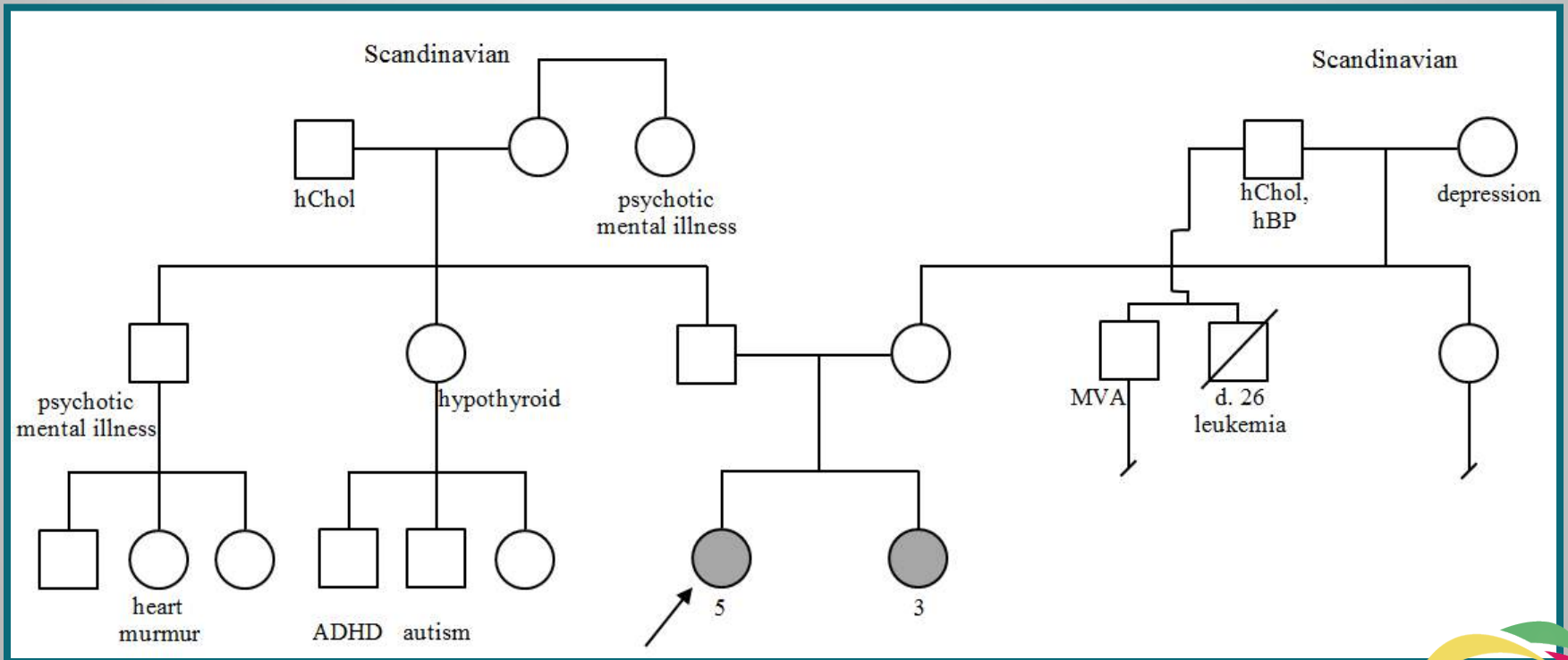
# Genetic Counseling



# Recurrence Risks



# Genetics...it's a Family Affair



# Rare & Undiagnosed Diseases as an Island

“living like this is like having your house be on fire and lacking the tools to put it out”

“I don’t feel like I can explain what’s happening to medical professionals or that they always believe me”

“helps just to know a program like this exists – we’re not the only ones out there”



# Social Uncertainty

- ✓ Feelings of isolation
- ✓ Lack of validation of concerns
- ✓ Don't have the words to describe experiences



# Social Support is **Everything**

- ✓ Finding someone with the same condition
- ✓ Finding someone going through the same things as you
- ✓ Finding someone who believes you



# Genetic Counselors' Role

- ✓ Identify sources of social support
- ✓ Provide vocabulary
- ✓ Connect families with each other and support groups





# Shared Goals

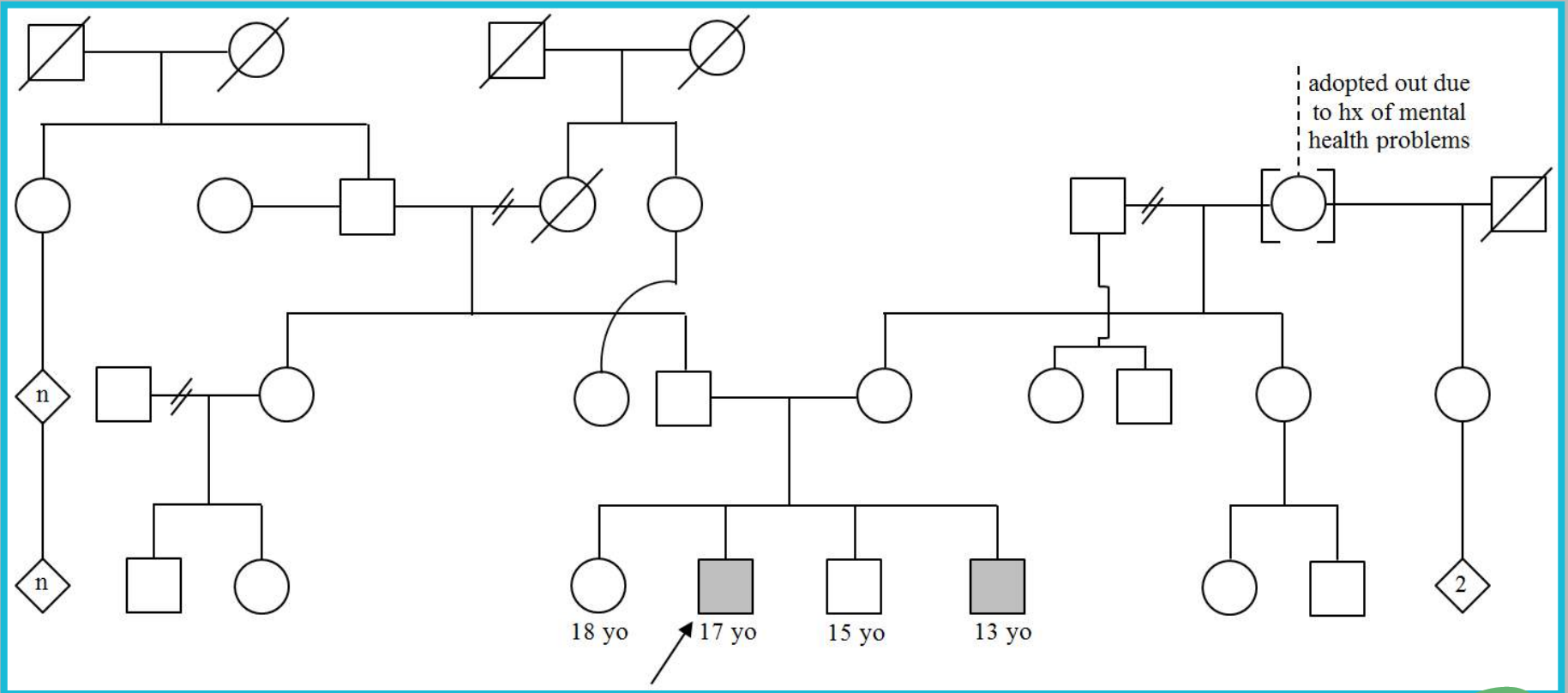
✓ Patient organizations

✓ Providers

✓ Researchers



# A Lesson from the UDP



# Clinical Description

## 17 year old

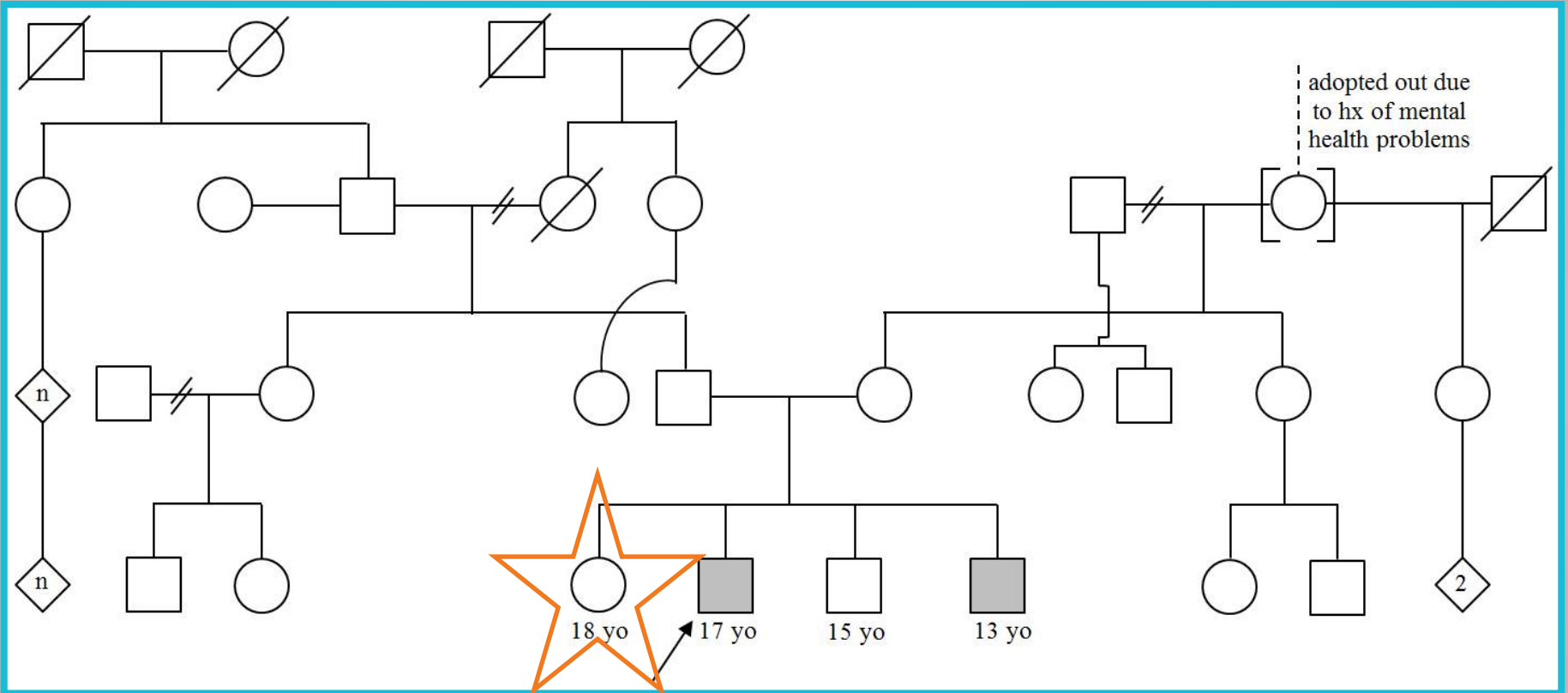
- Early onset schizophrenia at 3-4 years old
- Neurodegenerative course at 12 years old
- Long history of antipsychotics and steroids

## 13 year old

- Severe anxiety
- ADHD, bipolar disorder
- Cognitive impairment
- Neurological dysfunction



# “MK”



# Take Home Message

- What is Genetic Counseling?
- How can we help families with CDGs?
- Lessons I've learned



# Thank you!



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**Foundation Glycosylation (FoG) is the official sponsor of the videos targeted to the  
“SSIEM Official Satellite Symposia – Second World Conference on  
Congenital Disorders of Glycosylation (CDG): a challenging story of sugar trees”:**



The Foundation Glycosylation (FoG) founded by Duncan Webster (Canada), is the official sponsor of the videos of all oral session that will be given during the conference.

This material will be available in the Youtube channel dedicated to “SSIEM Official Satellite Symposia – Second World Conference on Congenital Disorders of Glycosylation (CDG): a challenging story of sugar trees” at:

For more information about the work of this organization which is focused on research to ALG9 -CDG (CDG -1L), visit the following link: <http://www.thefog.ca/main.html>

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