



Demystifying Genetic Counselling: An exploration into genetic counselling for rare diseases

**EURORDIS Open Academy Meetup:
January 2024**

Sophia Cross – UK NHS Trainee Genetic Counsellor

Introduction: Sophia Cross



- Final year trainee genetic counsellor
- Working at Guy's Hospital, London, UK
- Training through the NHS scientist training programme (STP) a 3 year programme combining work based training and a part time MSc in clinical science
- BSc Biology, University of Warwick, UK
- MPhil Genomic Medicine, University of Cambridge, UK
- MSc Clinical Science (Genomic Counselling), University of Manchester, UK

Introduction: Ali Kay

- Rare disease parent & researcher
- Patient advocate (muscular dystrophy)
- Research Fellow in Centre for Personalised Medicine, University of Oxford

- EURORDIS Summer School in Medicines R&D
- DPhil (PhD), University of Oxford, UK
- MSc Genetic & Genomic Counselling, Cardiff University, UK
- MSc Psychology, University of Central Lancashire, UK

Quick poll: What do you think about genetic counselling currently?



What is genetic counselling?



“ Genetic counselling 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 Counsellors in the US (NSGC, 2006)

The aims of genetic counselling are to help the individual or family to:

- ✓ Understand the information about the genetic condition
- ✓ Appreciate the inheritance pattern and risk of recurrence
- ✓ Understand the options available
- ✓ Make decisions appropriate to their personal and family situation
- ✓ Make the best possible adjustment to the disorder or risk.

Elements of genetic counselling

1. Information gathering

- Patient “agenda”, medical history and family history.

2. Information giving (education)

- Inheritance, condition, risk assessment, testing options, management, prevention, support, research

3. Counselling and psychological support

- Help patients adapt to their situation and choices, and issues that stem from the condition

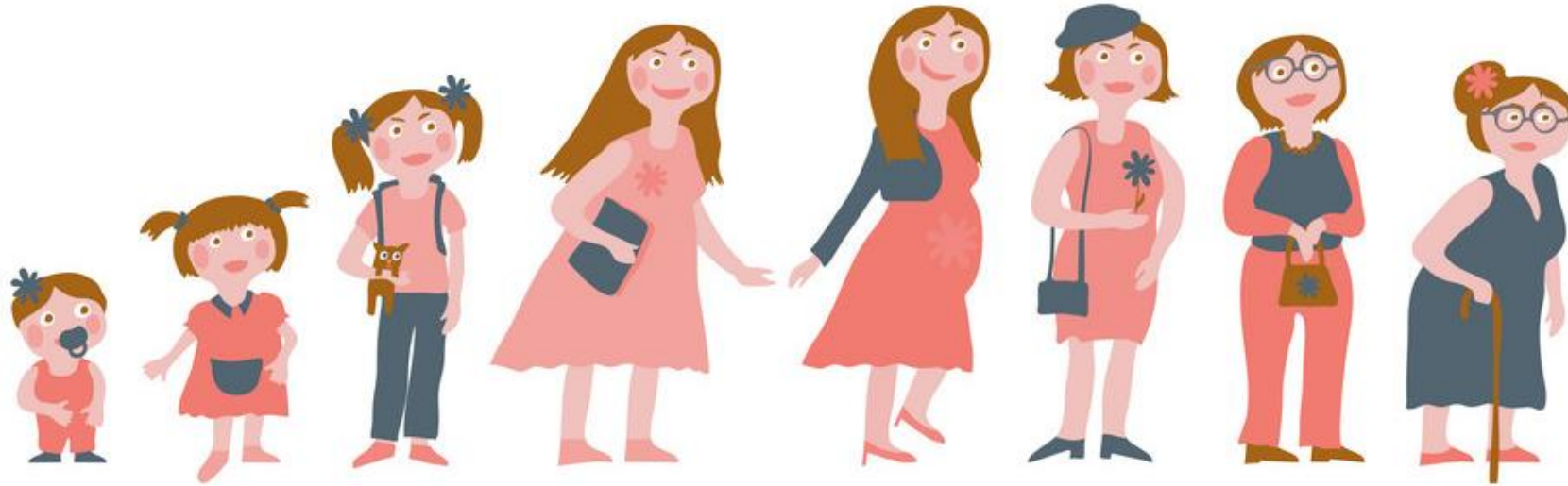
Common questions in genetic counselling

1. Why do I need a genetic counsellor?
2. Condition related: *"What is it?"*
3. Risk and genetic testing: *"Have I got it?"*
4. Inheritance: *"Can I pass it on?"*
5. Emotions: *"Did I do something to cause this?"*
6. Screening/management: *"What can I do to stay alive for my children?"*

1. Why do I need a genetic counsellor?

Adulthood

- Diagnosis
- Management advice



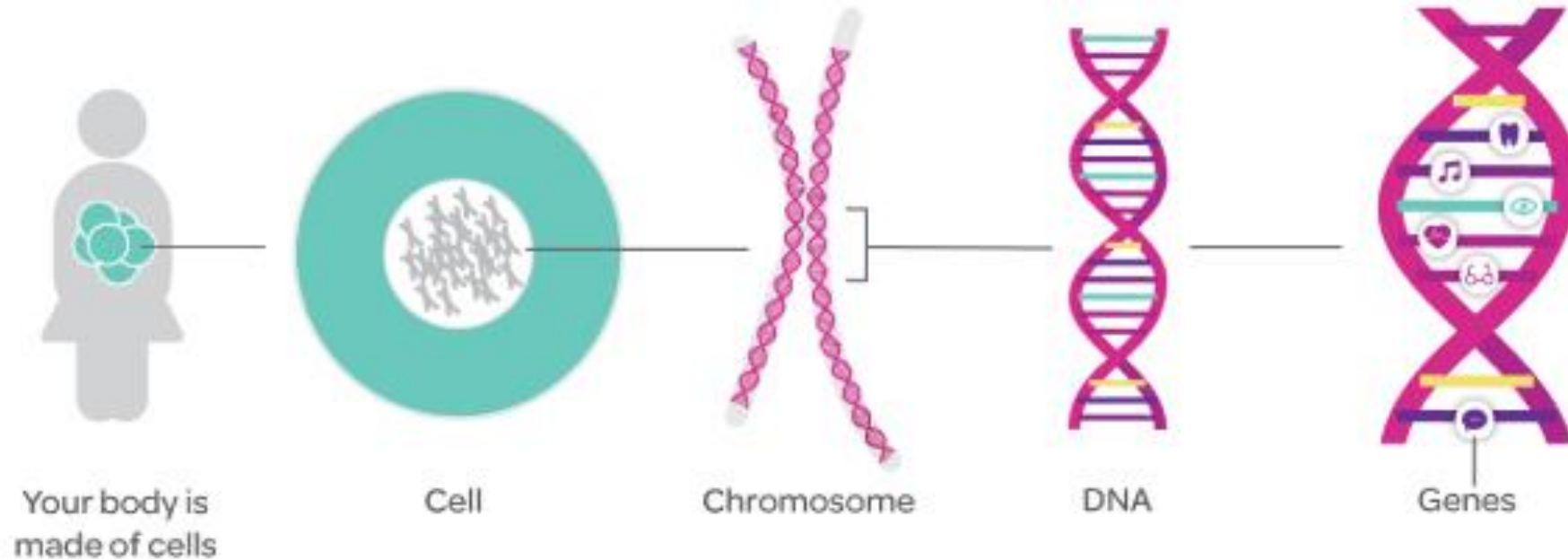
Childhood

- Diagnosis
- Newborn screening

Adulthood (Pre-conception)

- Discussion of options

2. Condition related: “What is it?”



Genetic disorders can be:

- **Chromosomal:** Affecting the chromosomes. E.g. missing or have duplicated chromosome material
- **Single-gene (monogenic):** Occurs from a single gene mutation

3. Risk and genetic testing: *"Have I got it?"*

We communicate risk and chance to patients regularly.

A risk assessment is based on: The condition, The patient and The Guidance

Why?

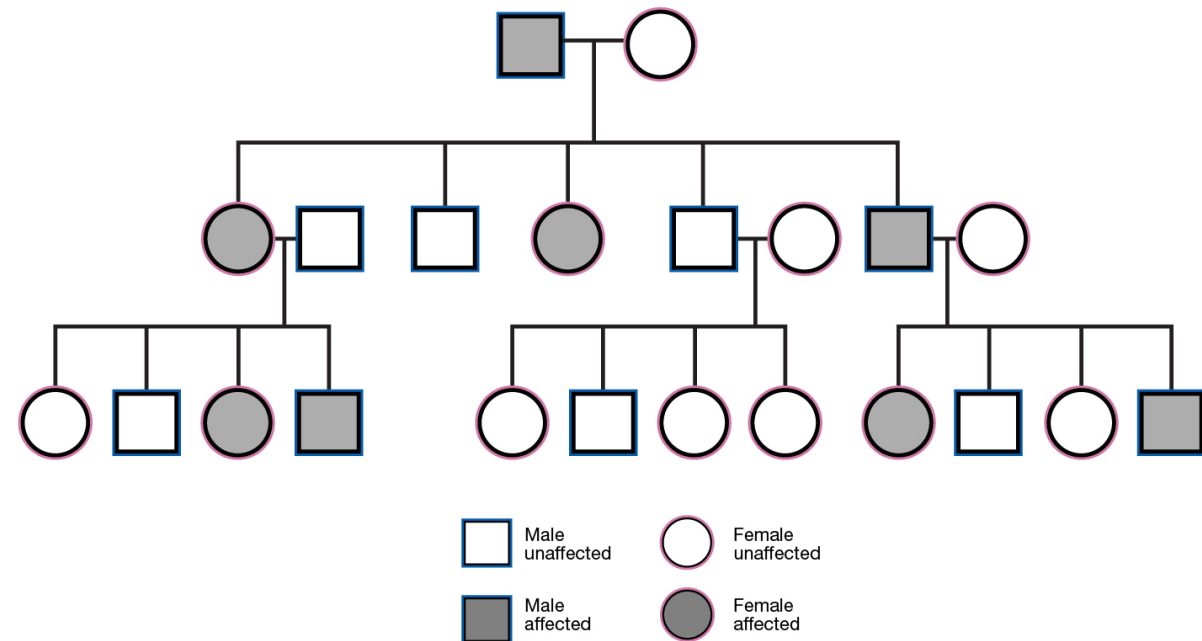
- To help patients understand factual knowledge
- To help patients make informed decisions

What influences risk perception

- How risk is presented
- Influence of family and friends
- Life stage
- Lived experience
- Bereavement

4. Inheritance: “Can I pass it on?”

- Autosomal dominant
- Autosomal recessive
- X-linked dominant
- X-linked recessive
- *De novo*
- Germline mosaicism
- Mitochondrial



5. Emotions: *"Did I do something to cause this?"*

Natural emotions we explore

- Anger
- Denial
- Disbelief
- Grief & Mourning
- Adjusting to/coping with change
- Avoiding emotions
- Blame

6. Screening/management: *"What can I do to stay alive for my children?"*

- Screening
 - Cancer screening
 - Tracking disease progression or improvements
- Treatment options
 - Medication
 - Gene therapy
 - Dietary changes
- Symptom awareness
 - E.g. for cancer conditions where no screening exists
- Surgery options
 - Prophylactic surgery
 - To treat symptoms e.g. bone marrow transplant

The genetic counselling appointment

- Appointments last 45 minutes to 1 hour
- Face-to-face, virtual or telephone
- Pre-test and post-test genetic counselling
- In genetics we are part of many disease specialisms:

Cancer (Oncology)

Kidney (Renal)

Brain (Neurology)

Eye (Ophthalmology)

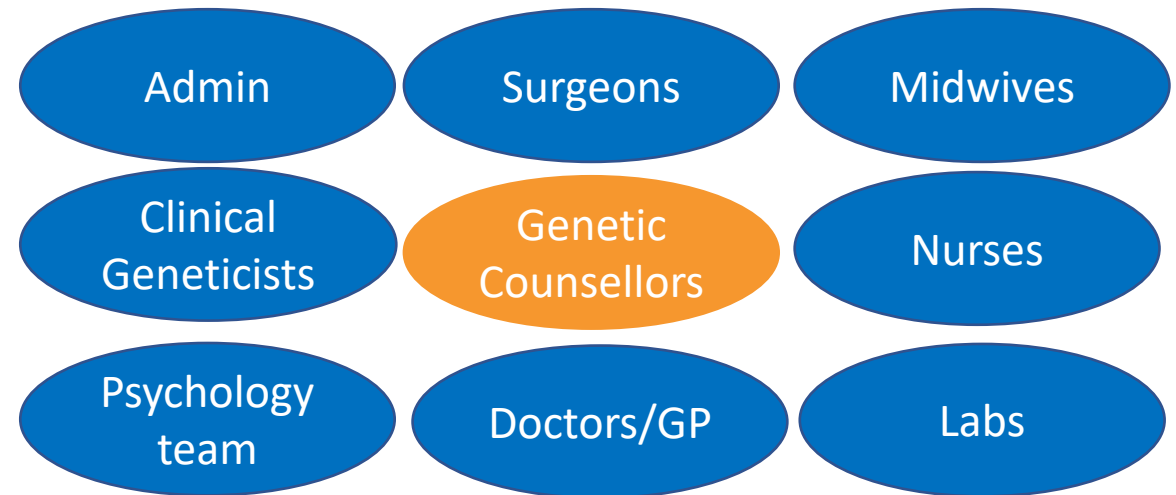
Metabolism
(Metabolic)

Heart (Cardiology)

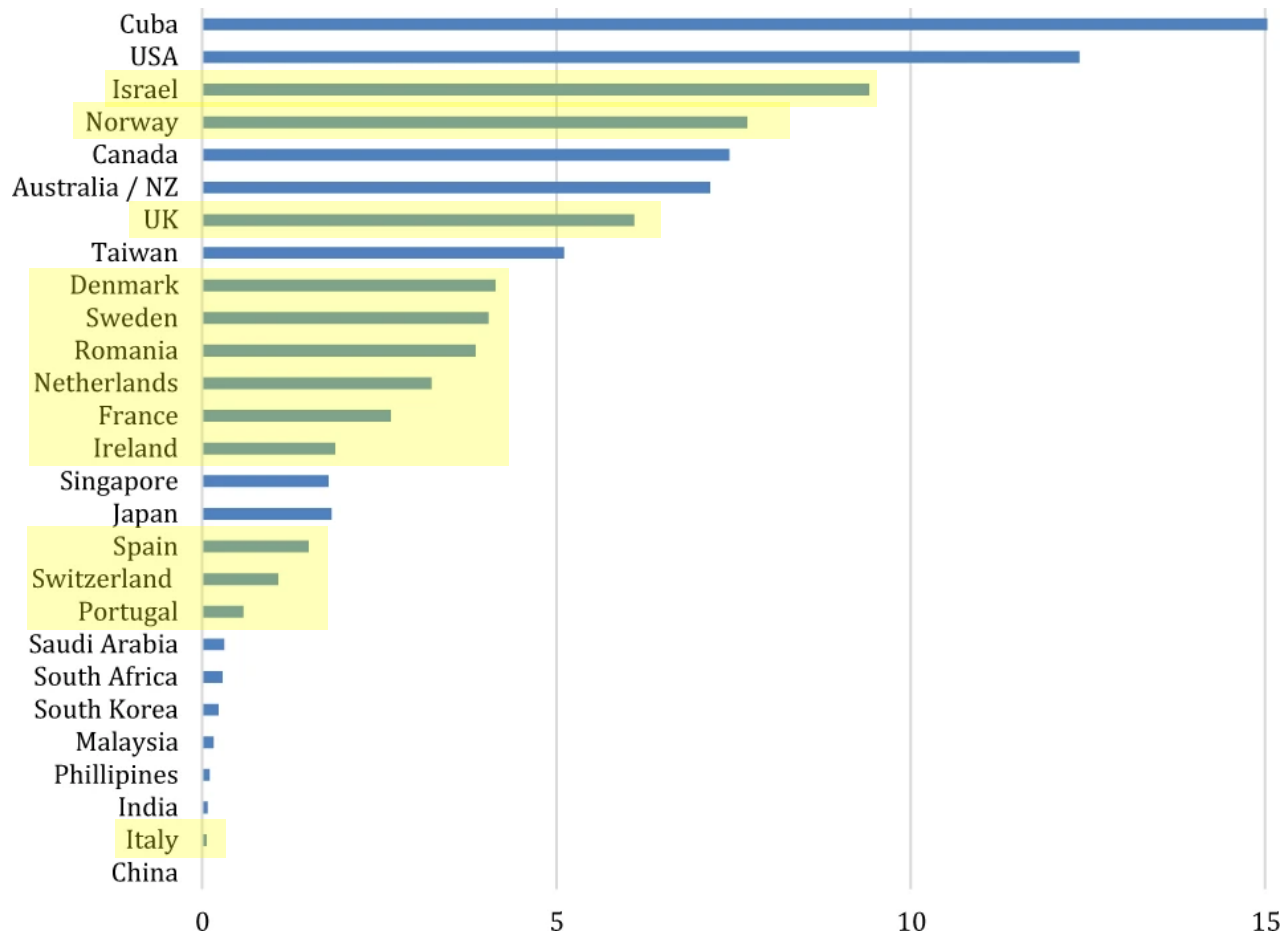
And many more...

Pregnancy (Prenatal)

Genetic counsellors do not work alone



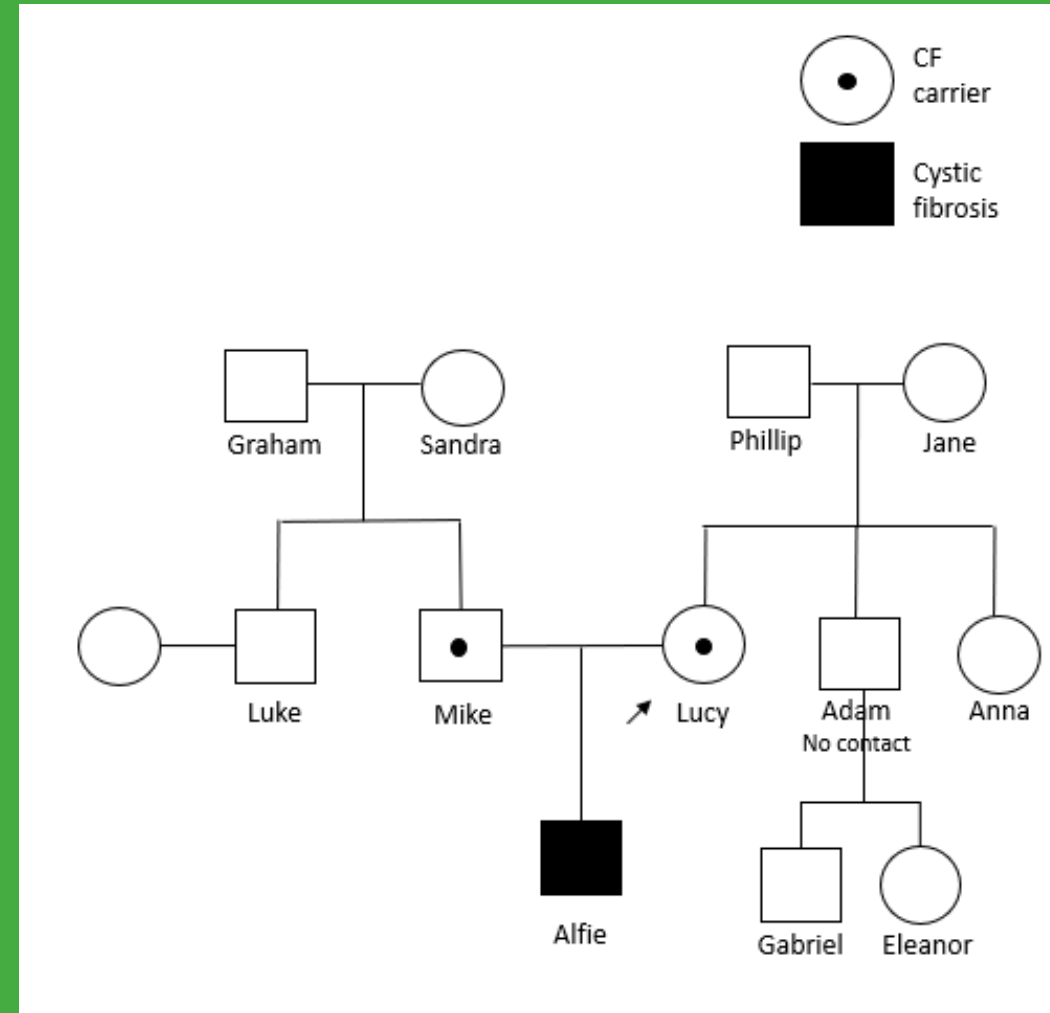
The status of genetic counselling in Europe



- Different professions can provide genetic counselling
- European countries vary in the availability and scope of genetic counselling
- GCs are found through national health-care services and by referral from health-care providers
- In other European countries where GCs do not provide clinical services, roles are varied
 - Laboratory work
 - Private companies,
 - Involved in healthcare provider education

Case example role play

- Mike and Lucy are referred to a genetic counsellor after their son, Alfie was diagnosed with Cystic Fibrosis (CF) on Newborn screening
- Neither Mike or Lucy's family have any history of CF, they would like to understand how Alfie has the condition
- We have taken a family history (right), we know that Mike and Lucy have no medical history of concern
- **We will discuss the condition, inheritance and implications for other family members**



Group discussion

- What did you think about the role play?
- Was this what you expected a genetic counselling conversation to look like?
- What did you think about the non-disclosure?

In summary

- Genetic counselling addresses the needs of individuals and families
- **Meet needs by asking what they are**
- Long term involvement with families due to impact on subsequent generations
- The emotional wellbeing of patients has to be considered alongside the genetics of the condition
- Ethical issues will arise and continue to challenge clinicians



Questions, thoughts, comments?

Thank you for listening.

Further reading

- List of European genetics societies or genetic counsellor societies

List of National Human Genetic Societies

collapse all

- ▶ All-Ukrainian Genetics Association Experts of Medical and Laboratory Genetics
- ▶ Armenian Society of Human Genetics
- ▶ Austrian Society of Human Genetics
- ▶ Belgian Society of Human Genetics
- ▶ British Society of Genetic Medicine
- ▶ Bulgarian Society of Human Genetics
- ▶ Clinical Genetics Society of Croatia
- ▶ Croatian Society of Human Genetics
- ▶ Cyprus Society of Human Genetics
- ▶ Czech Society of Medical Genetics
- ▶ Danish Society of Medical Genetics
- ▶ Dutch Society of Human Genetics
- ▶ Dutch Society for Laboratory Specialist Clinical Genetics
- ▶ Dutch Society of Clinical Genetics
- ▶ Estonian Society of Human Genetics
- ▶ Finnish Society of Medical Genetics
- ▶ French Association of Genetic Counsellors (AFCG)
- ▶ French Federation of Human Genetics
- ▶ French Society of Human Genetics
- ▶ Genetic Association in Bosnia and Herzegovina (Udruženje geneticara u BiH – GENUBIH)
- ▶ Genetics Society of Israel
- ▶ Georgian Society of Medical Genetics and Epigenetics
- ▶ German Society of Human Genetics
- ▶ Hellenic Association of Medical Geneticists
- ▶ Hungarian Society of Human Genetics
- ▶ Icelandic Human Genetics Society
- ▶ Irish Society of Human Genetics
- ▶ Israeli Society of Medical Genetics
- ▶ Italian Society of Human Genetics
- ▶ Latvian Association of Human Genetics
- ▶ Latvian Society of Medical Genetics
- ▶ Lithuanian Society of Human Genetics
- ▶ Macedonian Society of Human Genetics
- ▶ Malta College of Pathologists, Human Genetics Group
- ▶ Medical Genetics Society of Israel
- ▶ National Board of Clinical Geneticists of Georgia
- ▶ Norwegian Society of Human Genetics (NSHG)
- ▶ Norwegian Society of Medical Genetics
- ▶ Polish Society of Human Genetics
- ▶ Portuguese Society of Human Genetics
- ▶ Romanian Society of Human Genetics
- ▶ Russian Society of Medical Genetics
- ▶ Serbian Genetic Society, Section for Medical Genetics
- ▶ Slovak Society of Medical Genetics
- ▶ Slovenian Association of Medical Genetics
- ▶ Slovenian Society of Human Genetics
- ▶ Spanish Association of Human Genetics
- ▶ Swedish Society of Medical Genetics
- ▶ Swiss Association of Genetic Counsellors
- ▶ Swiss Society of Medical Genetics
- ▶ Turkish Society of Medical Genetics

(Slide in appendix in case pedigree needs changing)

