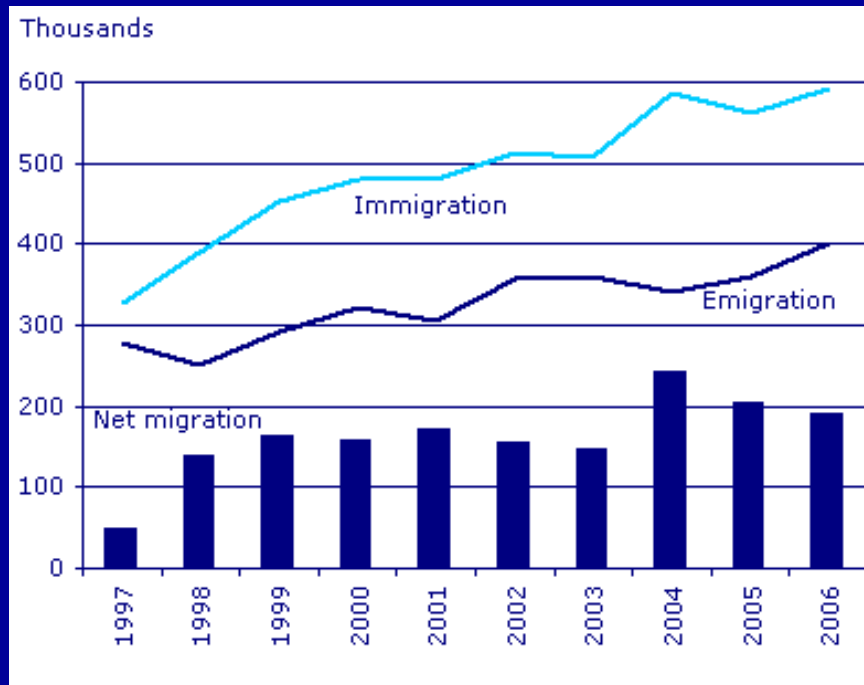


The New Rare Disease Challenges of Patient Migration and EU Enlargement

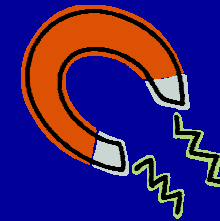
Jill Clayton-Smith
NW Regional Genetic Centre
Manchester, UK

Migration and the EU



PUSH

Unemployment
Natural disasters
Lack of services
Poor housing
Poor farming
War



PULL

Jobs
Education
Healthcare
Housing
Safety

MPI DATA HUB

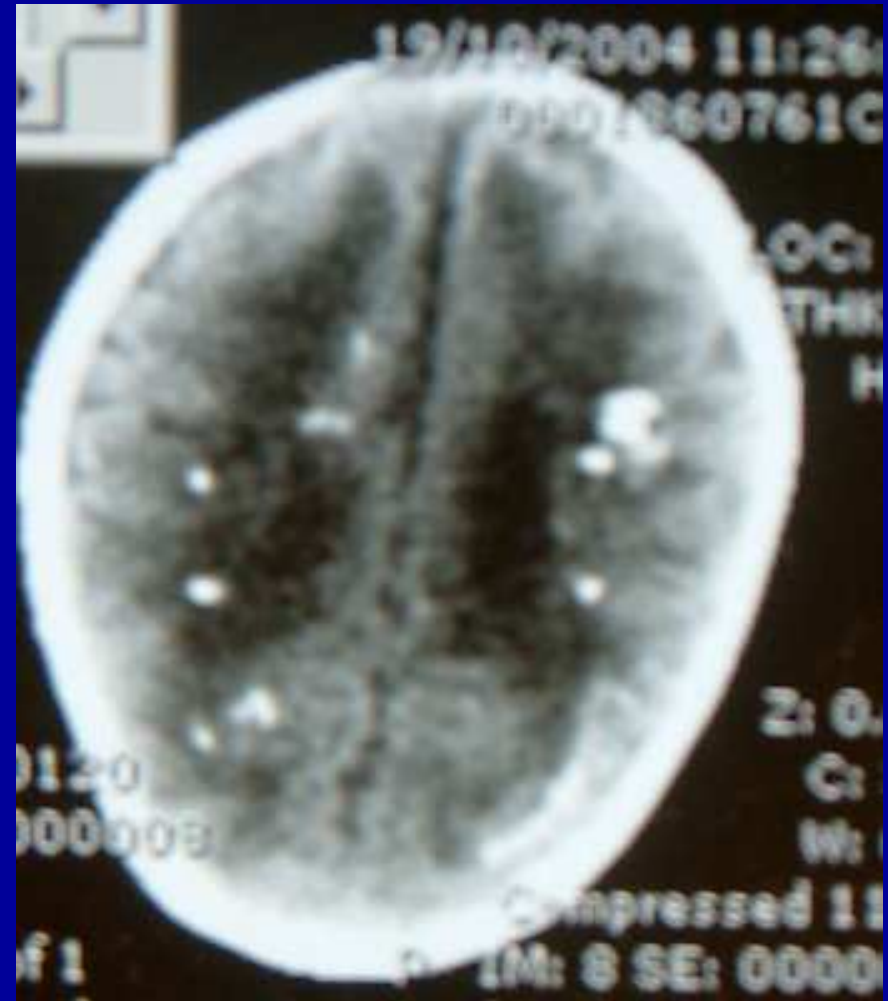
MIGRATION FACTS, STATS, AND MAPS

Migration and Health

- The majority of individuals migrate for economic reasons, to the mutual benefit of the migrant and the receiving country
- Migrants carry with them health “footprints”
- Often have a health profile associated with poverty
- Rare diseases form only a small proportion of the health burden of migrants, but have important implications as require specialist services

Challenges In The Clinic

- A young couple present with their first baby who is severely disabled. There was intrauterine growth retardation and a small head size. A scan has shown intracranial calcification



Two Possible Scenarios

- White, non-consanguineous couple
- Access to previous medical records of mother and baby
- No language barrier
- No significant cultural differences
- Consanguineous Turkish couple
- Previous medical records unavailable
- Language barrier
- Need to be aware of cultural issues

Diagnosis:
Congenital Infection

Diagnosis:
Aicardi-Goutière Syndrome
(autosomal recessive)

What Are The Challenges?

- The likely diagnosis may vary depending on the background of the family
- Not having access to previous medical records can make things difficult
- Language barrier may cause problems
- Ability to work with interpreters, including family members
- Being sensitive to cultural differences
- Discussing reproductive issues

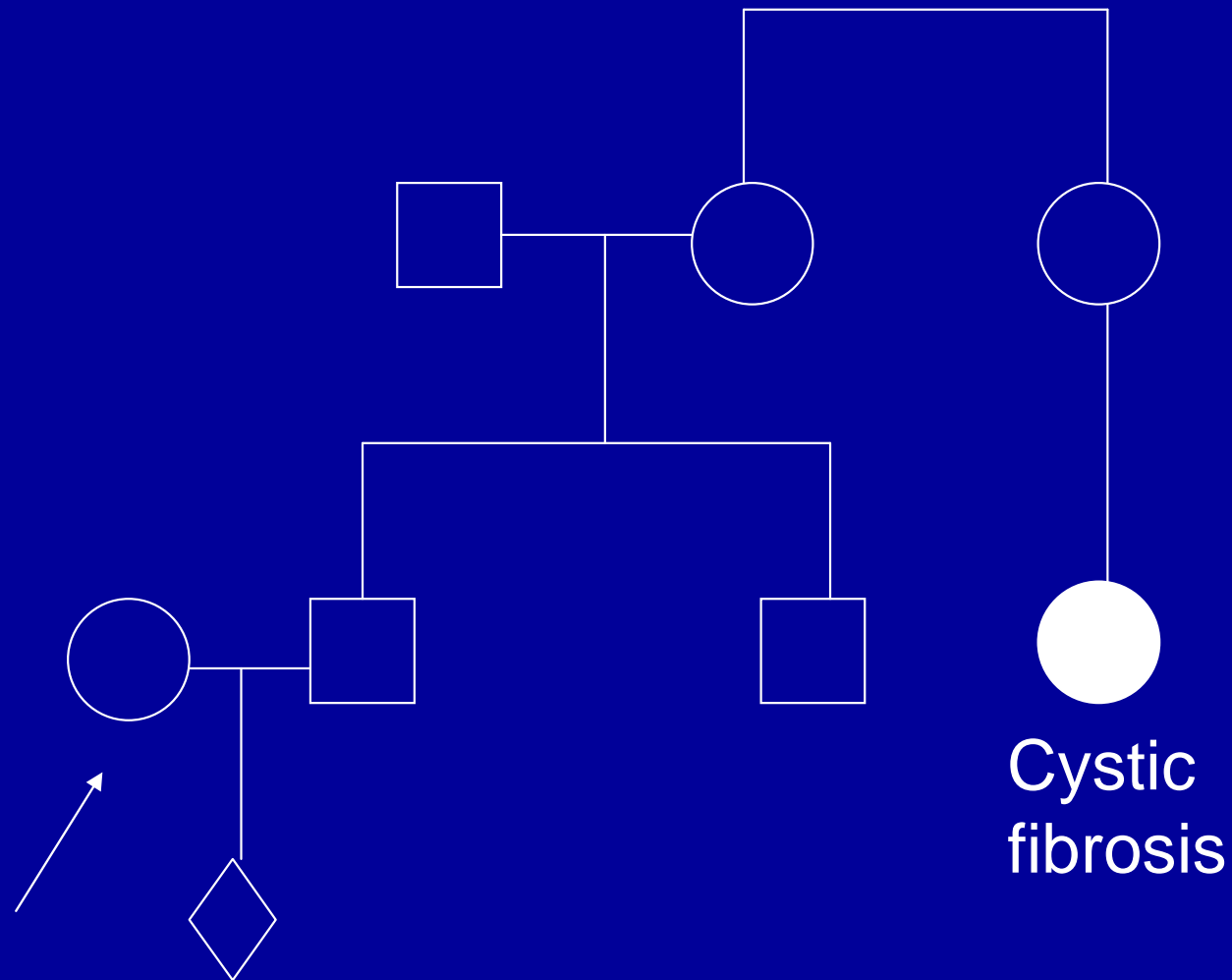
Consanguinity and Rare Diseases

- Rate of birth defects in 1st cousins 4-4.5% compared to 2-2.5% population risk*
- Major reason is autosomal recessive disease
- Social and cultural reasons for cousin marriages
- Aim to identify and counsel couples at high risk

% consanguineous		
Turkey	22	
Spain	6.8	(26)
British Pakistani	69	
White UK	0.4	
Scotland	2.6	

*Birmingham Birth Study Bundy et al.

Family 2; Cystic Fibrosis Carrier Testing in an Asian Family



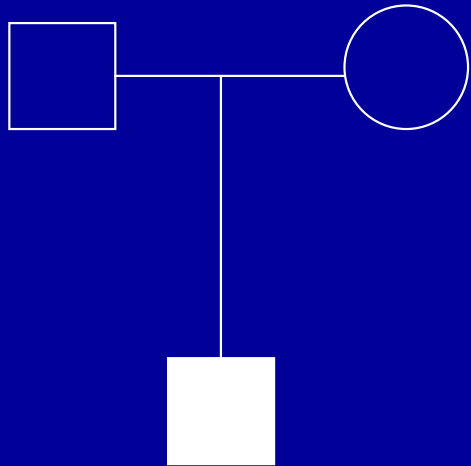
Issues Arising In Family 2

- Confirmation of diagnosis
- Any mutation analysis of affected individual?
- Which mutations to look for; laboratory need the correct information
- Long term outcome for children with CF depends on where they are managed
- Cascade screening will be difficult

Family 3; Floppy Baby

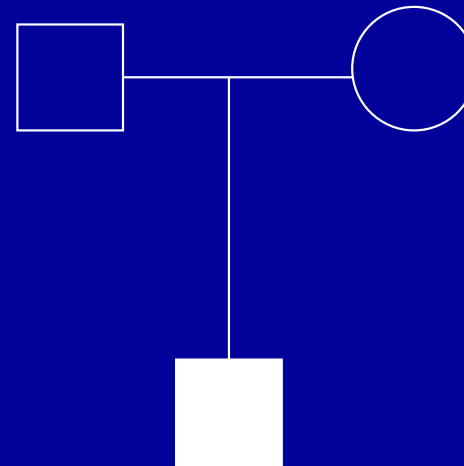
French

Italian



Roma

Roma



Spinal Muscular Atrophy
Type 1

Congenital Myaesthesia

Lessons From Family 3

- Some conditions are very rare: You have to do your homework
- Accessing specialist management expertise and testing may be difficult if the condition isn't common in the country where patients present for care
- Difficulties with providing support

Family 4: The Dysmorphic Child

- Dysmorphic features can be difficult to assess in different ethnic groups, could delay diagnosis
- Difficulties providing suitable literature and support may lead to isolation

How Can We Improve Things?

- Networks and Registers to promote knowledge transfer
- Address ethnicity and diversity issues, effective working with interpreters, facilitate access to language tuition for families
- Create specialist Interpreters, Genetic Counsellors and Link workers
- Extended role for rare disease groups
- Ensure access to accredited genetic testing

Arguments for targeted screening programmes?

e.g. Smith-Lemli-Opitz syndrome

EU Incidence overall 1 in 10-70,000

Czech republic 1 in 10,000

Slovakia 1 in 15,000

Poland 1 in 2000-1 in 3000

Conclusions

- Although health care is not the major reason for migration, migration has major implications for those managing rare diseases and for the patients concerned and needs to be taken into account when planning future healthcare services
- There are significant resource issues eg in provision of interpreters and translation
- Role of networks and patient groups is important
- There are also opportunities to share best practice and for collaborative research