Who needs to see a clinical geneticist?

Cambridge University Hospitals NHS
NHS Foundation Trust

Dr. Richard Sandford,
Academic Department of Medical Genetics and
Cambridge Renal Genetics Clinic,
Addenbrooke's Hospital.

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Specialist genetic services are particularly skilled in:

- Making or confirming the diagnosis of a genetic condition
- Calculation of the probability of inheriting a condition or of being a carrier
- Explanation of the genetic or inherited basis of the condition, and potential implications for family members
- Discussing and undertaking genetic testing and explaining results
- Giving up-to-date Information about a condition, its management and treatment

Genetic services are experts in dealing with families, often over several generations, and providing genetic expertise for any age group affected by, or at risk of, disorders in any body system.

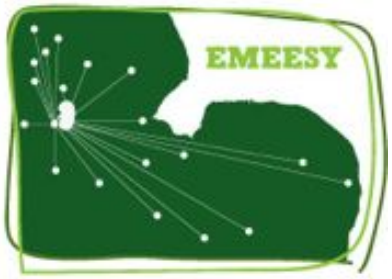
NHS National Genetics and Genomics Education Centre

East Anglian Medical Genetics Service

Clinical Genetics
Molecular Genetics
Cytogenetics

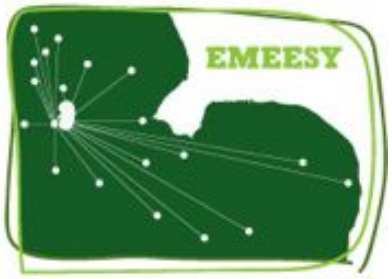
The majority of specialist genetic services are provided through Regional Genetic Centres which provide a co-ordinated clinical and laboratory service, often in close association with an academic university department. The clinical team usually includes Clinical Geneticists (medical doctors who increasingly may have subspecialty expertise), doctors in training, Genetic Counsellors and supporting administrative staff.

Services are delivered in **clinics** in the regional centre, in outreach clinics in district general hospitals, in ward or hospital department consultations and in visits to the families' homes. There is increasing provision for 'advice and guidance' referrals and telephone consultantions.



Whom should I refer?

- A person with a possible or known genetic condition in the family wanting to know if a diagnosis can be made in them or their offspring, and if so, the risks, options and treatment.
- A person with a family history of cancer, wanting to know if they are at increased risk, and if so what options they have.
- Parents of a child with multiple medical problems and/or learning difficulties, wanting an expert opinion particularly as to whether an underlying diagnosis can be made
- A person with a genetic condition wanting information about the condition and the management / surveillance options available for them
- A person with a family history of a condition undertaking a genetic test to determine if they have inherited the condition or are a carrier
- A couple considering pregnancy where there is a genetic condition in the family, requesting information about reproductive options
- A couple who have had recurrent pregnancy losses or children with multiple anomalies
- People requiring access to and explanation of complicated genetic tests



How is genetics relevant to my role?

- Do I see conditions with a genetic basis?
- Are some genetic conditions more common in my area?
- Can I take a multi-generational family history?
- Can I draw and interpret a multi-generational pedigree?
- Where do I find further information?
- Is genetic testing available and can I interpret the result?
- Can I identify at risk family members?
- How do I undertake family screening?
- Am I able to discuss genetic information with patients and their families?

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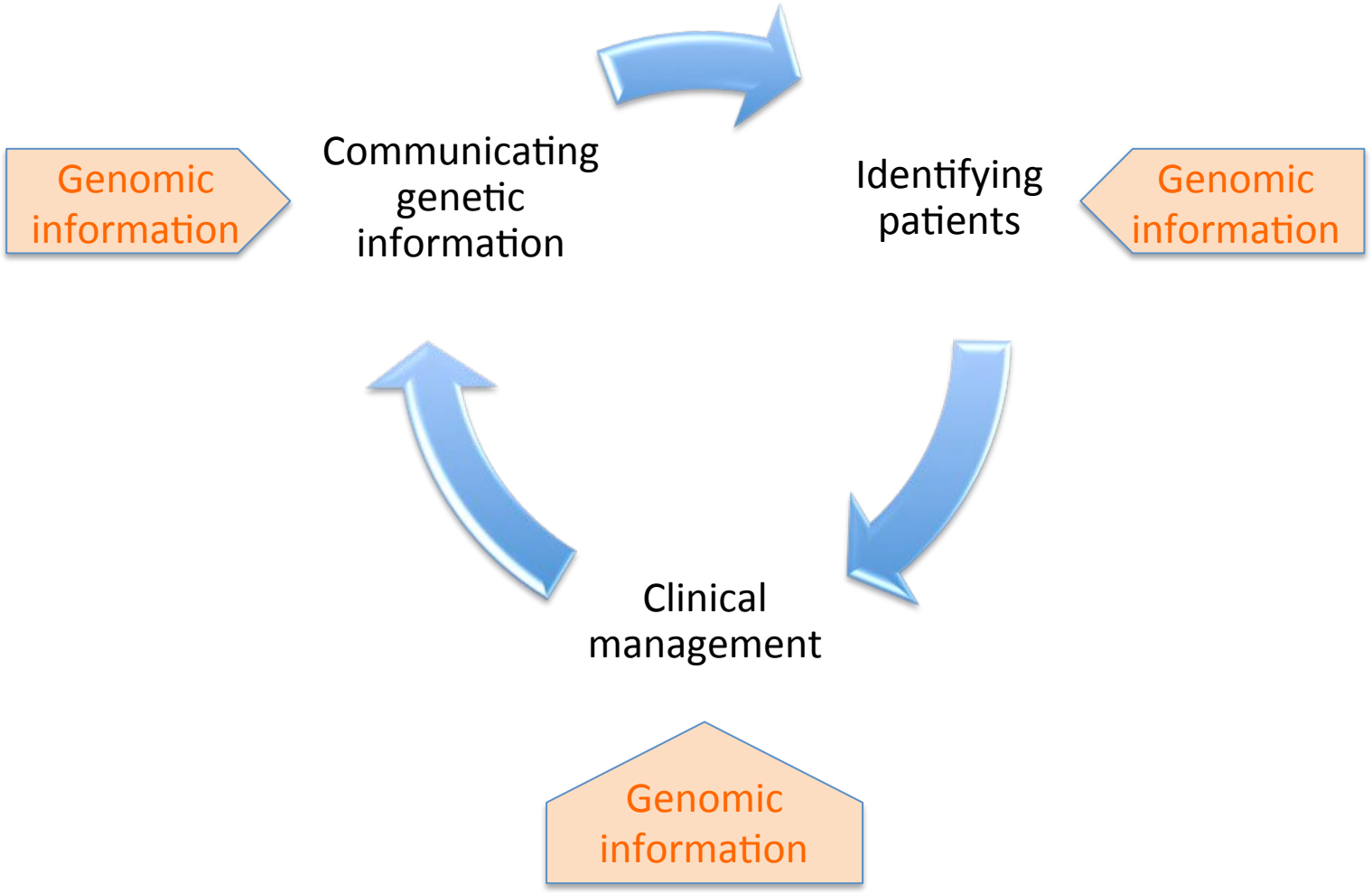
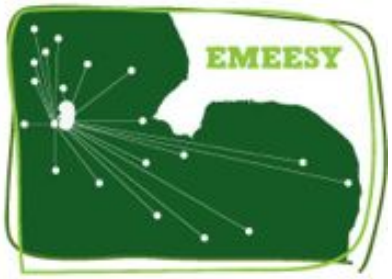
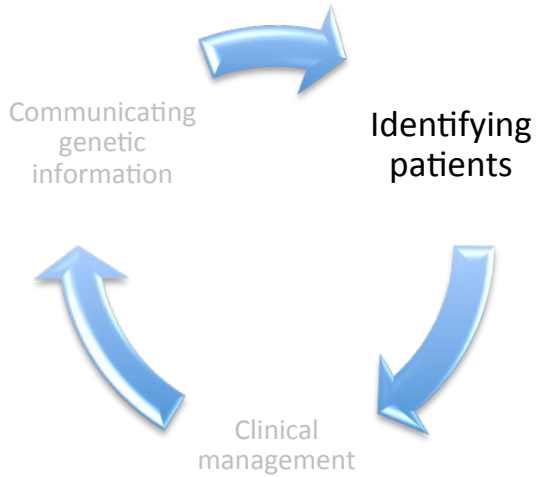
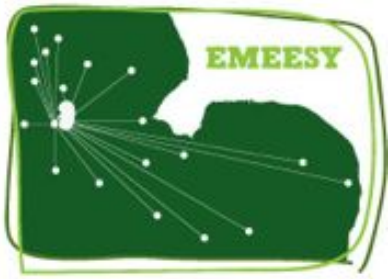
The Patient Pathway

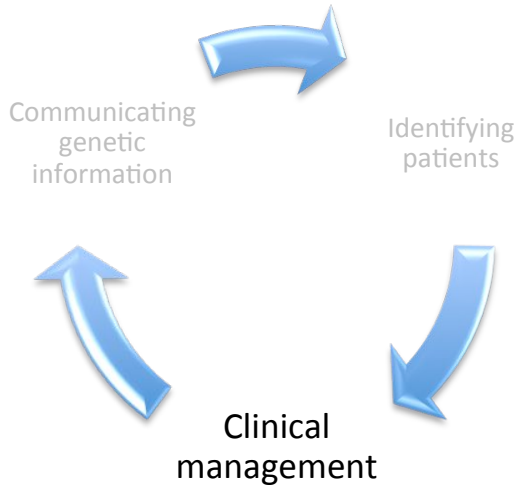




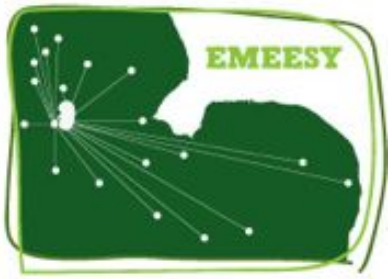


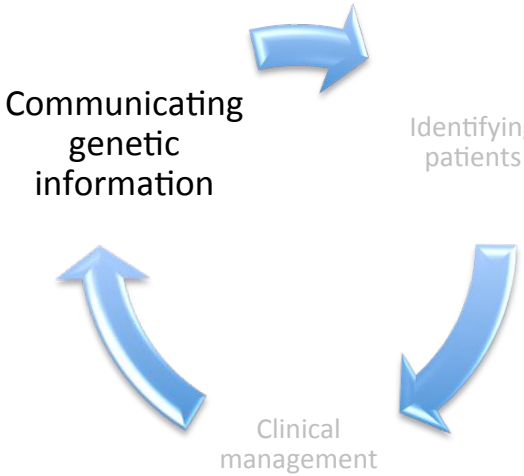
- Recognising the genetic contribution to common and rare diseases
- Recording relevant family history
- Drawing a pedigree
- Assessing genetic risk
- Performing diagnostic tests
- Specialist referral





- Genetic testing (germ-line and somatic)
- Information
- Referral to genetics services

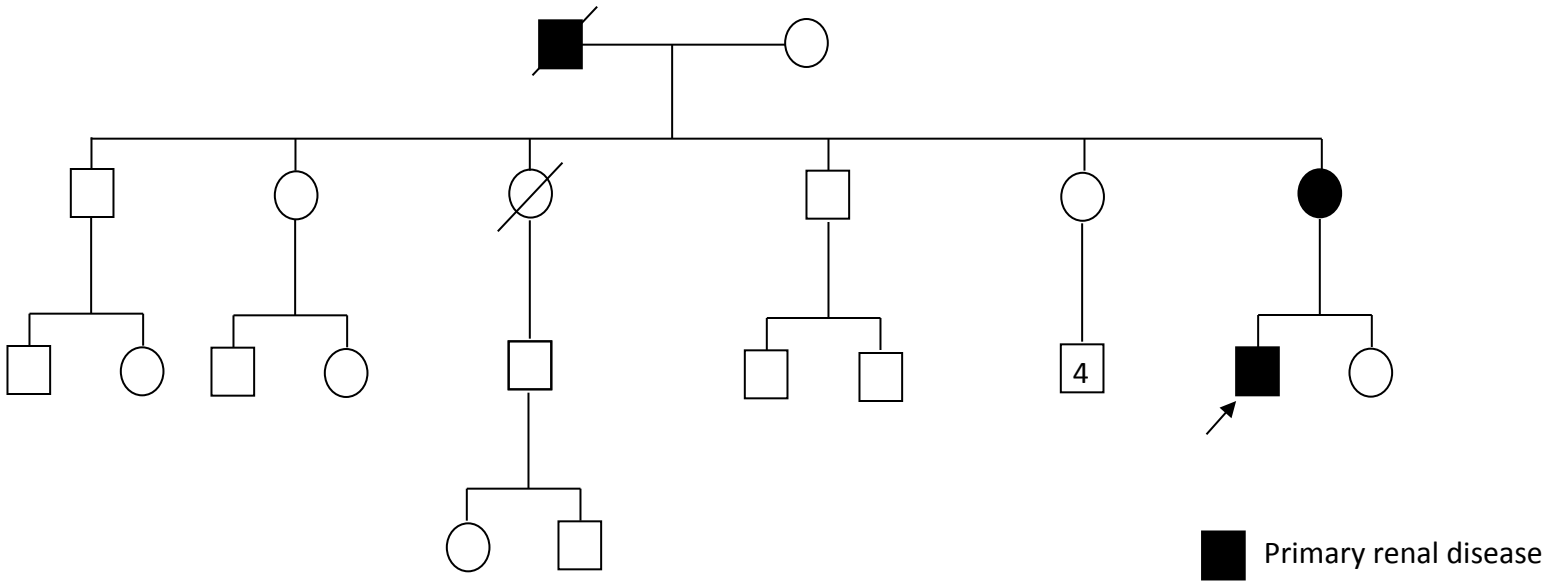


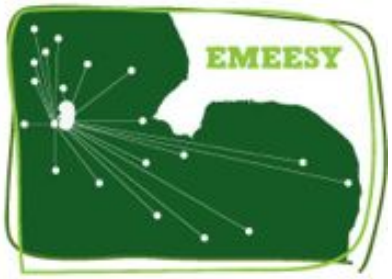


- Using appropriate terms
- Explaining inheritance
- Consent and confidentiality
- Sharing information
- Family screening
- Reproductive options

Scenarios

- A 14 year old boy was referred for investigation of high blood pressure after a visit to A and E for a sports related injury
- The BP was persistently elevated on a home BP monitor!
- The family history revealed that that the father had a long history of hypertension and a recent diagnosis of CKD stage 3
- The paternal Grandfather had died whist receiving haemodialysis





- Likely genetic diagnosis
- High genetic risk identified
- Renal biopsy performed in father
- Genetics referral

Identifying patients

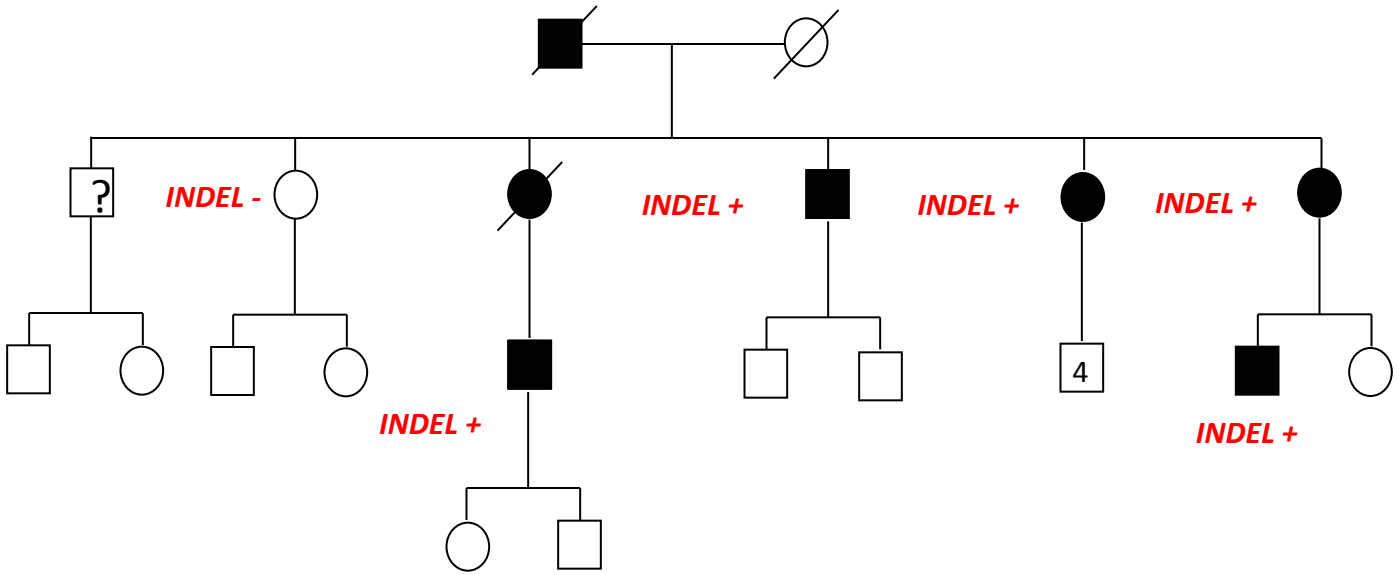
Communicating genetic information

- Family screening
- Predictive genetic testing
- PGD

Clinical management

- Gene panel testing
- Diagnosis UMOD nephropathy (FJHN, ADTKD)
- Hyperuricaemia
- Treatment
- Follow-up

Scenarios



Common referrals in Renal Genetics Clinic

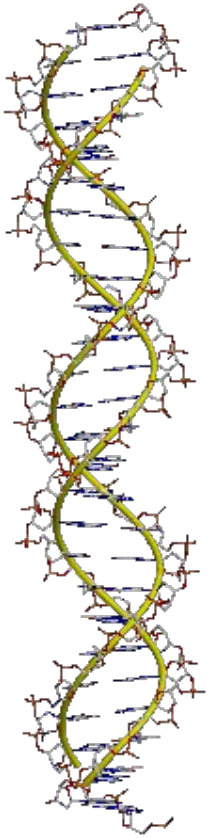
- Known genetic renal disease
- FH of renal disease-unknown primary diagnosis
- Genetic testing-diagnostic and predictive
- Renal cystic disease
- Evaluation of renal malformation syndromes
- Microscopic haematuria
- Donor evaluation-exclusion testing

Who needs to see a clinical geneticist?

- Obvious isn't it!
- Please feel free to discuss cases
- MDTs (pathology, radiology, genetics)
- Establish contacts with your local service to build up expertise
- Attend clinics

Resources

Many and varied!



http://www.geneclinics.org/
http://www.orphan.net/
http://www.ukgtn.nhs.uk/gtn/Home
http://www.geneticseducation.nhs.uk/
http://www.library.nhs.uk/genepool/
http://www.ncbi.nlm.nih.gov/
http://omim.org
http://www.ncbi.nlm.nih.gov/books/NBK1116/
https://www.renalradar.org

www.addenbrookes.org.uk/serv/clin/genetics/genetics_index.htm