What do nurses need to know about genetics?

In this article...

- How patients react to a genetic diagnosis
- Why health professionals need to understand the role of genetics in disease
- Referral to specialist genetic clinics

Author: Laura Boyes is consultant genetic counsellor, West Midlands Regional Genetics Service.

Abstract: Boyes L (2013) Rare diseases 6: what do nurses need to know about genetics? Nursing Times; 109: 49/50, 24-25. This sixth article in our series on rare diseases explores the impact of a genetic diagnosis on patients and their families. As genomic testing becomes mainstream, practitioners need to understand the role of genetics in disease and know how to refer patients for help and advice.

As genomic testing becomes mainstream, practitioners need to understand the role of genetics in disease and know how to refer patients for help and advice.

It's late on a Friday afternoon. A midwife calls. She has just given a couple the results of an amniocentesis test that indicates a complicated chromosome anomaly. Blood samples have been taken from the parents but the test results will not be available for a week or two.

In the meantime, the couple are plunged into a confusing world of uncertainty where they need to understand complicated genetic information and make difficult decisions about the pregnancy, while managing guilt that this may be inherited and the implications for relatives.

Dealing with a genetic diagnosis

A genetic diagnosis raises similar issues at any life stage. The “why me?” question and the lack of control can be hard to understand.

In my experience, it is human nature to rationalise the unexpected, and families often attribute blame and judgement to help re-establish control and reduce the threat. Questions patients may have after receiving a genetic diagnosis include:

- Is my condition inherited?
- Will my children get it?
- Will other members of my family get it?
- Can I have genetic testing?
- Is prenatal diagnosis available to help me avoid passing on the condition?
- What screening, treatment and prevention is available?

Feelings of guilt and shame are common, but rarely articulated. Close relatives can harbour differing experiences, beliefs and values, which shape their feelings and choices around genetic testing, which can create tensions.

Managing these sensitivities requires a delicate balance between respecting autonomy and facilitating decisions – for both the person and the family – while maintaining an empathic, non-directive and non-judgemental approach.

While many genetic conditions are rare, collectively they are common. They range from more common conditions such as cystic fibrosis to the rare and very rare conditions described in this series. People with rare conditions often experience isolation and can benefit greatly from the time and specialist skills of staff in genetics clinic, so identifying and referring these patients is important.

Genetic conditions can occur over several generations, but can also appear to occur spontaneously. Questioning about extended family members helps to identify patterns; several affected relatives, a young age of onset of a medical problem, or one person having several apparently unconnected conditions can all be indicators.

5 key points

1. A genetic diagnosis can cause confusion and uncertainty for patients
2. One in 10 people have a genetic component to their condition
3. Patients with rare conditions can feel isolated
4. Autosomal dominant conditions often affect individuals in every generation
5. Autosomal recessive conditions are more common where parents are closely related

Microarray DNA chip: a microarray test can carry out many genetic tests in parallel.
**What causes genetic conditions?**

Our genetic material, DNA, contains all the instructions for us to develop, grow and function. DNA is wrapped up tightly on 23 pairs of chromosomes. One copy of each chromosome in the pair is inherited from the mother, and one from the father.

Genetic conditions are caused by changes to these chromosomes or to the code of the genes carried on them; these are called mutations.

Most genes are carried on the autosomes, the chromosomes numbered 1-22, which are the same in men and women. Conditions caused by these genes can be inherited in a dominant or recessive way.

**Autosomal dominant conditions**

Autosomal dominant conditions often affect people of either sex in every generation because only one altered gene out of the pair is needed to cause the condition (Fig 1). Each child of someone with an autosomal dominant condition has a one in two (50%) chance of inheriting the altered gene and having the condition too.

**Autosomal recessive conditions**

An altered gene or chromosome from both parents is needed to cause an autosomal recessive condition. Where both parents are carriers, each child has a one in four (25%) chance of having the condition. Inheriting from only one parent means the child is a carrier and may pass it on to the next generation. Recessive conditions are difficult to recognise but are more common where parents are closely related, because the likelihood that they are carriers for the same condition is greater (Fig 2).

**Sex-linked conditions**

Each person has a pair of sex-specific chromosomes: an X and a Y chromosome in men, and two X chromosomes in women. Conditions caused by alterations in genes on these chromosomes usually appear only in one sex or the other.

Most sex-linked conditions are caused by alterations in genes on the X chromosome and affect almost entirely males. This is because males have only one X chromosome, while females with the same gene alteration have a second, normal copy of the gene on their other X chromosome so are healthy carriers.

A female carrier has a one in two chance of passing on the altered gene to each child, but daughters would be carriers: only sons would have the condition. Conditions inherited in this X-linked recessive way can sometimes be identified because only males in a family are affected.

**Referral**

Recognising families with these patterns and referring them to a regional genetic centre improves diagnosis, management of symptoms and risks, facilitates patient understanding, gives options for prevention in future generations, and may help with the development of treatments through research and gene therapy.

**The genomic revolution**

Traditionally, genetic testing was targeted at one or two genes associated with a condition. However, technology can now examine chromosomes in finer detail and multiple genes in parallel.

It may soon become cheaper to examine a whole genome as specific genes. Inevitably, this will increase complexity and uncertainty about the clinical meaning of results. However, it will make genetic diagnoses available to more people, more quickly and cheaply, and support specialties using genetic tests more routinely. As genomic testing becomes mainstream, all practitioners will need to understand genetics and its importance in disease. NT

**RESOURCES**

- British Society for Genetic Medicine: www.bsgm.org.uk/information-education/genetics-centres
- National Genetics and Genomics Education Centre: www.geneticseducation.nhs.uk

---

For more articles on genetics nursing, go to nursingtimes.net/genetics