There are now almost 2,000 tests for genetic diseases available, compared with around 500 just 10 years ago.

Advances in genetic and genomic research mean that identification of a genetic condition – or susceptibility to disease – is now possible in routine healthcare as well as being offered on a commercial basis, as highlighted in other articles in this series.

**Impact on family members**

Genetic tests have huge potential to guide the management of disease in an individual, but at the same time these tests may reveal that others in the family are at risk (see table below for examples). For instance, identifying that a woman has a BRCA1 gene mutation means that her sisters and daughters have a 50% chance that they have also inherited this condition. That figure is the same for her brothers and sons, although their risks of cancer will be lower. Affected relatives might benefit from the interventions available. The others can be reassured that they have not inherited the familial genetic risk.

**Genetic services and testing**

Genetic diagnoses are not new to medical practice and some indirect genetic tests – such as ultrasound for polycystic kidney disease – have been around for many years. Until recently, the routine delivery of predictive DNA tests has largely been the preserve of regional genetic services.

Unlike general practice, they often hold family rather than individual case notes and an initial consultation usually involves a detailed description of the family – people who might have an interest in subsequent diagnoses.

Most patients seek genetic advice with at least a partial aim of also helping their relatives. But there may be situations where family members are not in close contact, or because of concerns to protect people from distressing information, relatives may remain in ignorance.

Health professionals may then know about individuals to whom they could provide more accurate information, but concern about breaching the confidentiality of another prevents them from doing so.

This is not an unfamiliar scenario to GPs with many members of the same family on their lists. It’s not unusual to know that one relative does not want another to know aspects of their medical history.

Usually keeping such confidences is standard practice, but where this impedes optimal management of a relative, there may be reasons to share what might be regarded as familial information.

**Implications for general practice**

The best way to ensure family members can avail themselves of appropriate genetic

---

### Supporting you with CPD

1 CPD hour

Completing this module is worth a suggested 1 credit towards the 50 a year the RCGP recommends all GPs collect for appraisal and in preparation for revalidation.

**Go to www.pulsetoday.co.uk/cpd**