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One Gene or Two - Clinical Genetics within Modern Medicine Today

When I tell people I am a clinical genetics doctor they say, "Oh, that's a shame, do you miss not seeing the patients now?" Sadly this is how many people view genetics and the images portrayed by the media and artists like Jake and Dinos Chapman in the recent exhibition at the Saatchi Gallery help perpetuate this idea of genetic monsters.

Personally I see patients all the time, working closely with Cytogenetic and Molecular laboratories. Medical Genetics is a diagnostic and counselling service for individuals or families concerned about the suspected or confirmed diagnosis of a genetic disorder. This may include affected individuals or those concerned about their risk on the basis of their family history. Other common reasons for referrals are families with a history of reproductive loss or congenital abnormality and developmental or sensory disability.

Since Langdon Down first described Down Syndrome in 1865, we have learnt to recognise many more typical facial appearances across a variety of disorders. We describe new syndromes even now. Our understanding of genetics inheritance and DNA structure has evolved tremendously in a short time given that chromosomes where first described in the 1940's. We are now able to look at the substructure of chromosomes, in particular the tips of the chromosomes called telomeres. Mutations in these ends can be the cause of developmental delay in up to 8% of previously undiagnosed cases. We can sequence genes and identify small point mutations. However it remains a clinical diagnostic speciality since we need to know which disease gene to test.

Clinical Genetics is a service which offers clinical diagnosis, laboratory investigations, risk estimation and explanation of this to the family to enable them to decide on the most appropriate course of action and to understand how technology may benefit them. Establishing a genetic diagnosis or risk may also help colleagues in primary and secondary care to choose the most appropriate patterns of treatment and care.

Medical Genetics used to be mostly about trying to diagnose children and the various conditions they had but increasingly now we are looking at the genetics of adult conditions and other common later onset disorders. Family histories of cancer and in particular breast and or bowel cancer are good examples. We are even beginning to identify possible genes for schizophrenia, manic depression and dementia. Ultimately of course most every trait and disease that we have will have some underlying genetic basis, if not as its absolute cause, its likely that the susceptibility to a particular condition will be due to underlying genetic determinants.

So where do we go to with all of this genetic information? From a patient and family point of view access and delivery of information are extremely important elements of a clinical genetic service particularly given that genetic diagnoses and the potential risk to individuals, the possibility of impact on their life and occupation or the birth of an abnormal child are extremely stressful times and patients often wish to be seen quite urgently. There are many sources of information, not least of all the Internet, but this is not always accurate and often alarming. Families need clear, reliable and compassionate advice.

Clinical Genetics, as a speciality, is an important part of that information giving resource together with making the diagnosis and trying to identify what other problems are associated with the condition and what the future might hold. Furthermore Clinical Geneticists are often an excellent conduit for linking to both patient groups and researchers.

Ultimately medical genetics is about the patient and is trying to take us from the gene to the molecule to the organ and how that fits in with the body in general. Therein lies the continuing hope for the future.