Genctic counselling for patients with retinal dystrophies

This finding indirectly provided the first assignment for a genetic disorder - a type of congenital cataract - which was known to be linked to the Duffy locus. Over several decades the capabilities for assigning specific medical conditions to chromosomal locations has advanced from the traditional Giemsa stain and fluorescent in situ hybridisation technology through progressively finer detail in which today gene chips read by specialised software can analyse hundreds and even thousands of genes at a time rapidly picking up a single base pair change.

Today the fruits of such painstakingly won knowledge arrive as well marketed and neatly packaged gene diagnostic kits. Such tools of genetic diagnosis choose to revolutionise the way in which primary ophthalmic care is practised, presented and managed in the coming decades, said Dr Krawczynski.

A couple presenting in the clinic with a family history of retinitis pigmentosa may be advised on the specific mutation within the gene or genes involved. They may be given reliable figures relating to risk. They may be offered prenatal diagnosis and if one or either is beginning to show symptoms, they may be given up-to-the-minute information on experimental trials open to patient recruitment. Although such a scenario is within our grasp today, Dr Krawczynski’s studies illustrate that there may still be some way to go before such a streamlined “customer friendly” offering becomes standard practice.

EURETINA delegates learned that there are still a number of wrinkles in the idealised picture of how genetics and clinical ophthalmology might interact over the coming years. Dr Krawczynski’s study was based at the Unit of Genetic Counselling in Visual System Disorders at the Department of Medical Genetics, Poznan, University of Medical Sciences, Poland. Between 1999 and 2006 the Unit admitted 534 families, 266 of which were diagnosed with ophthalmic disorders as illustrated below:

The analysis of these families and the process of their counselling enabled Dr Krawczynski and colleagues to identify a number of observations from which they concluded that genetic counselling for patients with retinal disorders was highly specific. Genetic heterogeneity and the complex symptoms and investigations of patients created a need for significant cooperation with other specialists. From the study assessment, issues such as variable expression, possible phenocopies, genocopies and atypical cases often required clinical experience and more specialised investigations that might often be available in smaller clinical centres.

In addition, several patients and their families showed specific needs which, according to Dr Krawczynski, “are not usually observed in other groups of genetically determined conditions. For some patients, knowledge of the genetic risk is more important than their individual prognosis. Usually it is necessary to cope with parental feeling of blame, and sometimes psychological support is vital.”

Dr Krawczynski emphasised the importance of regular ophthalmic follow-up, even if there is no possible treatment, as well as referral to other specialists to diagnose and treat possible extracocular symptoms.

Dr Krawczynski proposed a model of genetic counselling for retinal dystrophies, primarily as a process that should enable the patient to comprehend the basic medical facts. Similar to any medical tool, genetic tests will need to be directed and interpreted responsibly with care and knowledge to insure no harm is done. To achieve this goal, practitioners will need a thorough knowledge of both the capabilities and most importantly the limitations of genetic testing. A deep foundation in technical issues will additionally require coupling with a compassionate understanding of good counselling practices.

In Dr Krawczynski’s view an ophthalmologist trained in medical genetics or a clinical geneticist trained in ophthalmology or cooperation of both specialisations represent a clear requirement if the field is to progress. He noted that a proportion of ophthalmologists appeared to experience problems with pedigree analysis, risk calculation and choosing an interpretation of genetic tests. However, clinical geneticists appeared to have a more significant problem with ophthalmological symptomatology and investigations. So while cooperation might provide an obvious solution, Dr Krawczynski commented that sometimes these two specialists speak two different languages, and it may be very difficult to facilitate cooperation.

In general it would appear much easier to train an ophthalmologist in clinical genetics than vice versa. Whether the case, it is clear that a tidal wave of new medical tools is fast approaching and in Darwinian fashion individual practitioners will have little choice but to adapt and evolve at a similar pace to the technology itself.