Langerhans Cell Histiocytosis: an orphan disease

There is no single agreed definition of a rare disease. The European Commission on Public Health defines rare diseases as those which are ‘life-threatening or chronically debilitating and which are of such low prevalence that special combined efforts are needed to address them’. The term ‘orphan’ has been used in association with rare diseases in order to emphasise their relative neglect in terms of professional awareness and available therapeutic resources to need to deal with them. ‘Low prevalence’ in this context refers to less 1 in 2,000. By this standard, Langerhans Cell Histiocytosis (LCH), the subjects of this month’s review is a rare disorder indeed with a presumed prevalence of 2–10 per million children and 1–2 per million adults. It results from clonal proliferation of Langerhans cells which are dendritic cells found in the dermis and lymph nodes. LCH belongs to the group of disorders known as the histiocytoses and has been known by a number of previous names including Leterer-Siwe Disease and Histiocytoses X; the current terminology of Langerhans Cell Histiocytosis having been agreed by the Histiocyte Society in the mid 1980s. The aetiology is not known and clinical manifestations vary according to the extent of the disease process. Two broad categories are recognised: single and multisystem LCH. The review paper considers how LCH is now considered to be a malignant rather than a reactive disease process and this is reflected by its current treatment. In early stages smoking cessation is considered to be of benefit while more advanced disease may be successfully managed by use of steroids or chemotherapy. Newer therapeutic possibilities include the use of such agents as clofarabine and thalidomide.

The study of rare disease is relevant for several reasons, not merely because of the impact of associated morbidity on affected individuals and their families. While individual disease may be rare, when considered collectively they are not uncommon. It is estimated that 30 million people have a rare disease in Europe alone. As one epidemiologist stated ‘rare diseases are common’ in that every clinician will have a number of patients with rare disorders. Many patients with rare diseases have experienced either delay in correct diagnosis and some of the disorders in this category are at least partly amenable to early intervention. Furthermore, enhanced understanding of the basic molecular pathology of very rare diseases often yields benefits for patients who have more common pathology.

Statins: are they effective and safe?

The media and scientific debate on the efficacy and safety of stains continues. To add to this ongoing discussion I draw your attention to yet another meta-analysis on the use of statins as agents designed to reduce cardiovascular morbidity and mortality. The journal has already published both original research and systematic reviews on this subject. The review by Mill et al. represents an updated meta-analysis of 75 randomised control trials (RCTs). The overall conclusion supports previous findings that statins significantly reduce the risk of cardiovascular disease events. There was no significant difference in impact of individual statins. Interestingly, this analysis found no significant increase in rhabdomylysis or cancer in those subjects who took statins. However, it was of some concern to note that 17 RCTs reported an increased risk of the development of incident diabetes. The authors conclude that substantial efforts should be undertaken to ensure adherence to statin therapy. As costs of the drug fall they argue for increased access by groups of patients currently excluded because of resource implications.

Sheehan’s Syndrome and Russell’s viper

Envenoming due to snake bite is a public health problem worldwide with an estimated incidence of
between 1–2 million episodes per year resulting in about 100,000 deaths. These figures are likely to represent an underestimate due to inadequate reporting mechanisms in many developing countries. The death rate due to snake bites varies across the world with the highest mortality occurring in South Asia. Many of the published accounts of this subject focus on management of the immediate complications post snake bite. The case history by Antonypillai et al. describes a longer term complication following envenoming by a species of Russell’s viper which resulted in Sheehan’s syndrome. The victim was bitten by this snake and was initially successfully treated with anti-venom. However he subsequently developed hypopituitarism. The case is interesting as it illustrates the myriad complication of snake bite. It highlights the need for follow up of patients who have been bitten by this particular snake and for a high index of suspicion for the presence of hypopituitarism which may be overlooked.

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