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Editorial

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# Rare diseases and expensive drugs

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Immunologists/allergists are a physician group that is familiar with caring for patients with rare diseases. Conditions that come under the umbrella of our specialty include various primary immunodeficiency diseases, mast cell disorders, various autoinflammatory diseases and diseases of the complement cascade including Hereditary Angioedema (HAE). A rare or orphan disease is defined by the Europeans as that which occurs in 5 people per 10,000 population. There are 6,000–8,000 known rare diseases with this number growing annually following new discoveries; 5–8% of the population will be affected by a rare disease, 80% of which have a genetic component. While these diseases have a low prevalence, they have a high degree of complexity in either their diagnosis, their management or both. Most are life-threatening or are chronically debilitating.

For the individual, these diseases carry special challenges; little is known about them; there is difficulty finding specialists who are knowledgeable; there are very few specific treatments available and when they are, they are very expensive and thus out of reach of the majority of patients.

HAE is one rare disease that has received a lot more attention in recent years, mainly because of the registration and marketing of new agents with which to manage the condition. HAE is a rare autosomal dominant disorder with an estimated prevalence of 1:50,000 that has been described in 3 forms; types 1, 2, and 3. Types 1 and 2 result from deficiency in functional C1 inhibitor (C1 INH), either from low absolute levels or production of a dysfunctional protein. In the absence of adequate levels of C1 INH, subcutaneous and submucosal oedema result from the uninhibited action of vasoactive mediators, of which bradykinin is considered the most important. HAE is characterised by recurrent oedema of the limbs, trunk, face, abdomen and sometimes genitals without urticaria, typically taking 24 hours to peak and resolving over 48–72 hours.

The most serious manifestation is laryngeal swelling, which was reported in older cohort studies to result in fatal asphyxiation in up to a third of patients.

Caring for patients with this condition within Asia Pacific countries poses special challenges. We have no accurate data on patient numbers in individual countries; patients face many obstacles with gaining an accurate diagnosis because of lack of

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recognition of the condition and, in many countries, lack of access to specific tests. There is stigma attached to the distortion of facial features that occurs with swelling attacks and there is a significant impact on quality of life and physical functioning among those with frequent attacks.

A number of treatments for HAE are now available including replacement treatments and pharmacological treatments such as a bradykinin receptor antagonist and kallikrein inhibitor.

Drugs for "orphan diseases" are particularly difficult and expensive to develop and have a high price tag when they eventually come to market as the pharmaceutical companies attempt to regain the costs of their investments. No individual patient is able to bear the cost of indefinite treatment so they will require a governmental approach to funding such treatments.

Bearing the costs of expensive treatments for a very small number of individuals is particularly problematic in countries where there remain major public health issues with communicable diseases where the investment of similar amounts of money required for a single patient's treatment would see large scale immunisation programmes completed.

However all patients have a legitimate expectation of treatment whether their condition is common or rare. Our societies need to find solutions that allow access to high cost drugs for rare diseases while not impeding funding for the very common communicable and noncommunicable diseases and for public health programmes that benefit much larger numbers of the population.

Ideally, pharmaceutical companies who discover and develop treatments for rare diseases need to be part of the solution that allows access to these drugs. Some see development of orphan drugs as part of their research effort and contribution to the greater good; others may enter cost sharing agreements with various governmental funding bodies.

With rare diseases, there are only a small number of patients available for clinical trials so by necessity there are significant limitations on the number of studies that are performed. This poses difficulties when attempts are made to register new products.

Government agencies can assist by facilitating and simplifying the registration process for orphan drugs. Sharing assessments for marketing authorisation would relieve some of the financial burden pharmaceutical companies face in the registration process.

Professional bodies such as the Asia Pacific Association of Allergy, Asthma and Clinical Immunology have a duty to critically evaluate the various guidelines for management and find the most acceptable recommendations for their own constituents. These may vary from what is considered best practice in the United States or Europe but may better meet the needs of our patients while balancing this against the pressures on the health dollar in countries where communicable diseases still extract a huge toll.


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