

Primary Immunodeficiencies – A worldwide public health priority

The proper treatment and early diagnosis of Primary Immunodeficiencies is a worldwide public health priority. It is estimated that there are around 150 types of primary immunodeficiencies. Although primary immunodeficiencies is a diverse group of diseases they share a similarity in that people affected by these conditions are subjected to recurrent infections if they are not treated properly, as their immune system is unable to protect them against foreign infections. Immunoglobulin replacement therapy is the treatment of choice for primary immunodeficiencies. Failure to treat people with primary immunodeficiencies properly leads to impaired quality of life and in most severe cases early death.

There are many rare disorders to deal with within the public health system of any nation. They are still nowadays too often ignored but ignoring them is an extremely costly way to handle the issue. Not treating these patients can be an additional burden to the economy or just result in poverty and increased ill-health. This is particularly relevant for people with primary immunodeficiencies. Unfortunately because symptoms of primary immunodeficiencies often manifest as common affections such pneumonia, bronchitis, or gastroenteritis, without proper knowledge about what causes these symptoms, many doctors focus on treating the symptoms rather than the cause. This leads to a huge waste of healthcare resources as the infections keep coming back and costly and prolonged hospital stays are needed. Proper treatment through immunoglobulin replacement therapy has been shown to significantly reduce episodes of infection, hospital stays and enable affected people to lead normal active professional lives.

Let us look at some concrete examples of what can happen when, say, a late onset condition such as so-called ‘common variable immunodeficiency’ (CVID) occurs in a person aged, say 26 years, who has previously been of good health – an active and productive member of society. Suddenly they become ill. Their family doctor will immediately treat the symptoms – possibly by the use of antibiotics. They are not effective and so the doctor will prescribe more, possibly stronger, antibiotics. In time the infection passes but this has been a striking experience – a strange and persistent infection in someone who was previously in good health. A few months later there is a recurrence of the infection – and again the family doctor prescribes antibiotics, this time starting with a stronger antibiotic. However, the doctor will be unlikely to think ‘Now why is this – we have a second severe infection in two months in this previously healthy patient?’ Indeed it has been known for doctors to conclude that the person may be mentally imbalanced and in need of some psychiatric support –

rather than think ‘Could this be connected to the patient’s immune system in some way?’

Immune systems are what keep us healthy and help us to recover from illness – some faster than others. We all know that there are things that we do that impair our immune response – not enough sleep, too much alcohol and so on!! – and so it becomes surprising that more doctors do not think that way. This is again why primary immunodeficiencies in particular are a major public health issue – awareness by those who ‘need to know’ is still appallingly low. If the average family doctor was just that bit more aware he would immediately refer our friend – who has had two severe bouts of infection at the age of 26 years after a lifetime of good health – to the local immunology services for a very cheap test that would show up straight away that this person has an impaired immune system. Such an appropriate diagnosis would lead to a much better quality of life for this person thanks to appropriate treatment but it would also lead to a more efficient management of healthcare resources.

The tragedy of today’s reality is that there are many such people in the community – people who have held good jobs, who have raised families – who suddenly become a drain in society and on the quality of life of their own families through their ill-health, their inability to work and the failure of their health system to diagnose their condition and then to manage it appropriately.

Fortunately, the situation is slowly improving as healthcare decision-makers are starting to be more aware of the issue. In Europe, the European Parliament’s Scientific and Technological Options Assessment (STOA) Panel held a workshop on 17 March 2004 on the issue of primary immunodeficiencies. In 2006, the European Commission sponsored a Consensus Conference which brought together more than 100 experts in clinical immunology, PID care, public health, genetics, EU/national ministries of health and agencies, academic centres, public health laboratories, industry, professional organisations and patient groups were brought together to identify and develop public health strategies for PIDs. Similar steps are taking place in other countries around the world, greatly helped and encouraged by the lead taken by the International Patient Organisation for Primary Immunodeficiencies with the European Consensus Conference and their successful campaign, along with the International Union of Immunological Societies, to re-instate immunoglobulin on the World Health Organization’s List of Essential Medicines.

Politically speaking, the snowball has started to roll for primary immunodeficiencies, and as it rolls down the slope, it will become bigger and stronger and people will start to see it better, as they should, because people with primary immunodeficiencies no longer should be ignored. We hope that this book will do just that: help raise awareness about this important issue for the sake of many patients in need.

José Luis Valverde and David Watters
Editors