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Hereditary diseases a bigger worry

IT is time to lessen emphasis on acquired/secondary immunodeficiencies (which include HIV infection) and increase it towards congenital and primary immunodeficiencies (PID). Care of HIV has done well, experiencing a drop of 50 per cent in 11 years, from 6,978 to 3,393 cases last year (as quoted by Health Minister Datuk Seri Dr S. Subramaniam in *New Sunday Times*, May 25).

Meanwhile, PID has risen 10-fold between 2007 and 2011 (cumulative frequency of 108) compared with before 2003. If the trend continues, it would be more than 1,000 by 2016, just four years before 2020, provided the human capital "patient-based immunologists" are increased concurrently. PID is often missed by the general specialists.

PID is a chronic, mainly hereditary disease that occurs in 1:5,000 to 1:1,200 per population (according to figures from the United States).

Elsewhere, with the lack of awareness, only a fraction are diagnosed. For example, in Europe only two per cent of cases are diagnosed and only 0.1 per cent in Africa. As such, there is a need to create awareness in almost all countries.

Malaysia, too, has its share of PID, with records by a leading researcher standing at 180 (1986-2013). Taking the European data, Malaysia would have at least 9,000 PID cases (higher than HIV prevalence in 2013).

How severe is it? The immune system is the body's defence against germs and other invaders. Through immune response, the immune system attacks organisms and substances that invade our body and cause manifestation of disease such as pneumonia (blood infection), abscesses, arthritis (joint inflammation), and even meningitis (infection of the central nervous system).

In PID, the defect of the immune system makes infectious disease more frequent, severe and difficult to treat. Untreated or late treatment would make a child very sick, leading to death. PID is not less severe than HIV. PID, being a hereditary disease, may affect other members of the family.

We will take only two examples of PID (from more than 200 types) to illustrate the gravity of the situation:

DEFICIENCY of antibodies; and,

DEFICIENCY of T cells (T Lymphocytes) combined with deficiency of antibodies (SCID, or severe combined immunodeficiencies).

Deficiency of antibodies requires replacement with infusion of commercial immunoglobulin.

SCID is treatable with bone marrow transplant (BMT).

Both are effective if treated early with good quality of life until adulthood. For SCID without BMT, the infant is unlikely to pass his or her first birthday.

Immunoglobulin replacement therapy for patients with antibody deficiency requires intravenous immunoglobulin (IV Ig)) to be infused regularly every three to four weeks for a lifetime. With frequent intravenous administration, venous access becomes difficult with "spoilt veins". This problem is not only in babies but also in obese adults. A subcutaneous (beneath the skin) preparation is available in developed countries but not yet available here. BMTs for PID patients are not easily accessible in Malaysia. Only the rare ones have had the privilege of BMT at the Institute of Paediatrics in Kuala Lumpur Hospital and University of Malaya Medical Centre.

PID is best treated by clinical immunologists followed by a specialist paediatrician or physician with training at clinical immunology centres.

There is a need for more immunologist sub-specialists for children and adults. Although in Malaysia, most PID cases are seen in children, they do survive into adults. There are three trained paediatric immunologists, but only one trained for adults, and all are in the Klang Valley.

Recognising "patient-based immunology" as a subspecialty, or even as an area of "interest specialty" as a start, cannot be delayed. Meanwhile, the death toll rises.

With almost 180 patients diagnosed and many more undiagnosed, the PID community faces a gloomy picture with severe morbidity and high mortality. Is it not the time to alleviate the suffering of PID patients?

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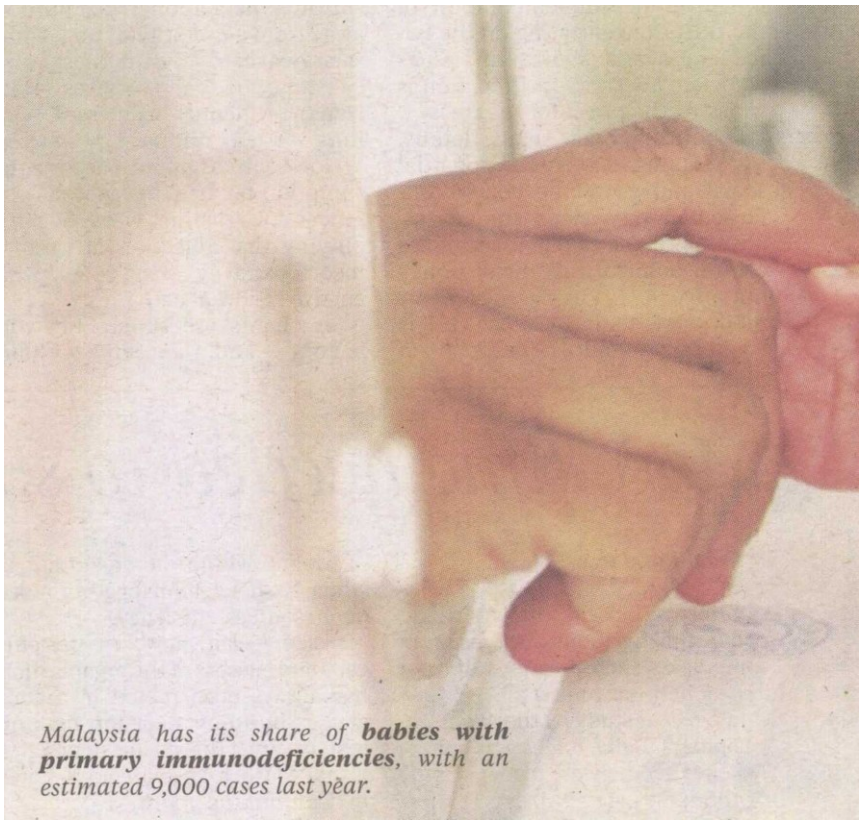

Malaysia has its share of ***babies with primary immunodeficiencies****, with an estimated 9,000 cases last year.*