24th April, 2018

This is World Primary Immune Deficiency (PID) awareness week. UKPIPS is highlighting the lack of awareness of PIDs within the GP community in the UK. Primary Immune Deficiency is not taught in any depth during initial Dr education. If they remember anything, they remember about Severe Combined Immune Deficiency, which results in very young children dying unless Bone Marrow Transfer is offered. However, the reality is that there are more adults diagnosed with a PID, (genetic diseases for the most part –either caused by a single gene mutation or by a combination of 'susceptibility' genes and unknown environmental influences), than there are children. The most common form of PID is where the person is unable to produce enough antibodies to counteract infections. We call all types of PID that affect antibody production Primary Antibody Deficiency (PAD). Just to make life a little more confusing, the most common form of PAD is called Common (the most prevalent) Variable (how it affects people can vary) Immune Deficiency (a lack of part or parts of the immune system) and we call this CVID.

So, whilst all of these acronyms can be very confusing, the bottom line is that people cannot catch a primary immune deficiency, nor can they pass it on to other people unless it is to their children by genetic transfer.

People typically live with CVID for approximately SEVEN years before they are diagnosed and during this time they will be living twilight lives, bouncing from infection to infection. Taking more and more courses of antibiotics, which may or may not work. The type of infection that undiagnosed CVID patients tend to get are those of the upper respiratory tract, the gut and the skin. THIS is where the difficulty comes in trying to get a diagnosis, because these will often be "normal" infections, that many people will get from time to time, such as sinusitis, chest infections, throat and ear infections, diarrhoea, infected eczema and so on. The difference is in the amount of infections and how difficult it is to clear them despite appropriate antibiotic therapy.
The more infections people have, the more scarring they get and once their organs are damaged, this can never be repaired. Many people end up with severely scarred sinuses, hearing damage and or bronchiectasis and this makes keeping them well, even when they are getting good replacement therapy, exceedingly challenging.

UK Primary Immune-deficiency Patient Support
Bretforton Suite De Montfort House 7e Enterprise Way Vale Business Park Evesham Worcestershire WR11 1GU
T: 01386 421228 **M:** 07531 076409 **E:** info@ukpips.org.uk **W:** www.ukpips.org.uk

This is SO frustrating, because, for many people, the answer can be found in a simple blood test that GP's are able to order. That is, to look at their immunoglobulin levels. However, not only is it the case that many GP's simply do not order these tests, but that if they do order them it is not unusual for UKPIPS community members to have been told that their GP "did not believe the test results because they said that PID was too rare for me to have".

Until and unless GP's begin to understand the reality that CVID and other PAD's can and frequently do develop in teenage years and at any time in an adult's life, including in old age, people will continue to have their lives ruined.

More information can be found on how to diagnose a PAD in the UKPIPS leaflet here.