

Frequently Asked Questions

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Answers to your questions about Primary Immunodeficiencies – now referred to as Inborn Errors of Immunity (IEIs)

Q: *If you or your child were diagnosed with PID/IEI what would be the first questions you would ask?*

A: Why did I get this? Can it be cured/treated? What treatment is available? What is the expected quality of life with this condition? What is the life expectancy? Could it have been prevented?

Q: *Can Primary Immune Deficiency/IEI be cured?*

A: Certain forms of Severe Combined Immune Deficiency (SCID) and an increasing number of IEIs, e.g. Chronic Granulomatous Disease can be cured by a successful stem cell transplant (e.g. bone marrow, cord blood, peripheral harvested stem cells). Therefore early diagnosis is important before irreversible organ damage has occurred. Gene therapy holds great promise for curing PID's, but later onset of cancers is still a problem. The more common antibody deficiencies are usually treatable with Immunoglobulin replacement infusions assisted by antibiotics and vaccinations.

Q: *How safe is the local immunoglobulin therapy in terms of transfer of infections e.g. Hepatitis and AIDS?*

A: Locally available Immunoglobulin (Ig) is as safe as any overseas product because the safety and quality controls, including nucleic acid amplification (NAT) testing for Hepatitis C and HIV are as strict as those for overseas products. In South Africa plasma is sourced from the blood donations of healthy unpaid volunteers. However it is a blood derived product and very rarely virus contamination (not HIV) has occurred overseas. Therefore the product, like any other transfusion should only be used for the correct medical indications and judiciously to be available for PID patients who depend on a regular supply.

Q: *How safe is the long-term use of immunoglobulin therapy?*

A: The safety profile of long-term Immunoglobulin therapy has been well established. All blood derived products are registered with the Medicines Control Council of South Africa in terms of their safety, efficacy and quality. There has been no documentation of residual effects with long-term use of intravenous immunoglobulins. Follow up of patients who have been on regular IVIG for decades now shows improved quality of life and equivalent ability to work compared to a person without PID. But treatment should be supervised by a doctor who regularly sees the patient holistically. Too little Ig treatment and excessively long intervals between treatment (generally more than three weeks in the case of intravenous Ig) can also result in recurrent infections and the outcome can be almost as bad as without treatment. This is not a complication of the treatment but of incorrect treatment and it is avoided by regular serum trough levels of IgG and medical check ups. As Ig transfusions do not prevent all infections, the IEI patient may also need to be antibiotics in addition, or require additional nutrition.

Q: *Primary Immune Deficiency is a chronic condition. Has the state established the infrastructure for home therapy e.g. home nurses, like in other countries?*

A: There is currently only assistance or plan for home therapy with certain Medical Aids. A universal access to home therapy would need extensive training and resources.

Q: *Could the medical fraternity advise a standard procedure in terms of clinical management of anyone with PID, obviously tailored to the need of the individual? Could this be made available on the website or in the brochure going to the doctors?*

A: Yes. Standard guidelines for diagnosis and investigations are available e.g. JMF guidelines – and the IDF (see useful websites). Treatment is often very individual, other than the standard guidelines for Immunoglobulin replacement. Specific medical protocols for individual diagnoses are available. In 2022 extensive recommendations for the indications and use of Immune replacement therapy for patients in South Africa have been agreed on by practitioners in the field in South Africa and published on the ALLSA website.

Q: *Is it safe for patients with Primary Immunodeficiency to receive the seasonal flu vaccine?*

A: The flu vaccine, now in stock in many pharmacies (although some stocks are already said to be running low) and to be made available in government clinics from April, is not “live” and therefore safe for PID patients who do not have severe T-cell deficiencies. Although it could be useful for PID patients’ immunity to the flu virus, it

is advisable to consult the patient's physician before having the vaccine. Before receiving the vaccine, the PID patient should also be in good health. If younger than two and a half, two vaccines will be required, four weeks apart. Preventative measures like hand washing should nonetheless continue throughout the flu season.

Q: *Is it necessary that the Polygam goes at 100ml per hour or can one speed up the by perhaps 150ml per hour and then 200ml per hour after a patient received Polygam for more than say 2 years?*

A: In practice experience is that the overall Infusion speed after the first 15 to 30 minutes (30 to 60 ml/ hour in adults and 0.01-0.02ml/ kg/minute in children) after the first three infusions can be advanced carefully, but if any side effects especially flushing or rash occur – step back immediately to previously tolerated dose. Depending on the individual patient and the diagnosis we have witnessed some very 'non standard' infusion speeds! If the patient has own IgG antibodies – like most Common Variable or Hypogammas do, we would not push the limits past any slight discomfort and not go past 150ml /hour infusion (120-150ml/hour) speed, (and children 0.08ml/kg/min) especially with home infusions. Use a 20 drop/ml infusion set. Also some patients are premedicated with Phenergan (antiallergy) and or steroids (Solucortef) and in this context one would also be careful – slower infusions may then not need these medications. Special caution for patients with kidney problems is advised. **All patients must be well pre-hydrated** – this may help prevent a number of the above side effects.

Q: *Bearing in mind patient confidentiality, is there a national registry of patients with PID (as they have in other countries) and if not, would the medical profession like one developed?*

A: A national registry with support from the industry has been established in 2006 and is stored on a secure server at Tygerberg Hospital, University of Stellenbosch. Only patient data with full patient/parent consent are stored (registered study with ethics number) and all data is strictly confidential. Information exchange of registry results with other countries and continents enables us to see different genetic patterns of diseases and incidences. It may be possible in future to assess the outcome on specific PIDs with good record keeping. This will result in better patient care, while serving to increase awareness for these rarer conditions.

Q: *Is genetic diagnostic available in South Africa?*

A: Yes. Please consult your physician/paediatrician for guidance on this in consultation with a genetic counsellor