

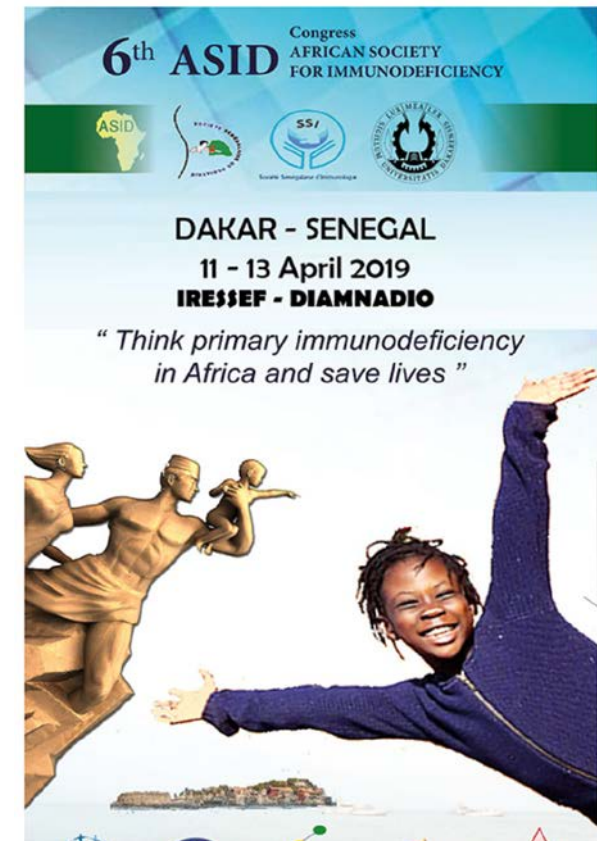
ASID DAKAR IPOPI 12 April 2019

What do I need to know about my condition?
(Frequently asked questions – or - What Questions to ask)

Que dois-je savoir à propos de ma maladie ?
(Questions les plus fréquentes)



Monika Esser
NHLS Immunology Unit Tygerberg
Stellenbosch University, South Africa



If only we
listen.....

our patients
will ask

Ask.

- Be direct.
- But also listen, ask follow-up questions
- Ask for clarification
- Do not be afraid to ask what others deem as 'dumb' questions. NO Questions are dumb!

The Invisible Mentor

JANUARY 26, 2019 BY [AVIL BECKFORD](#)



- The appropriateness and relevance of the answers you receive depend on the questions that you ask.
- **A question is the vehicle** to find out the answer to something – it is a way **for you to further your knowledge and gain factual information.**
- American novelist, Thomas Berger once said, “The art and science of asking questions is the source of all knowledge.” **Learn how to formulate and ask** intelligent questions.
- But your questions are also an opportunity to CAREFULLY educate your caretaker on the PID !

First Question : Whose “Fault” is it ?



IT IS NOT YOUR FAULT !

PID in the family is OUR Illness/Problem

- You don't "catch" PIDDs like the flu.
- You or your child were born with an abnormal gene that affects the immune system.
- Sometimes this PID runs in families (inherited). Or it may have just happened spontaneously (de novo) .
- White blood cells fight infections. With some PIDs some of these white blood cells are missing they don't function well.
- This makes one more likely to get sick from infections in PID.
- Nobody is being punished !

The time to diagnosis.....

.....
Why –what -
how

- The diagnosis of my/your PID
- Le diagnostic des DIP

- *“Dad and I are getting desperate for answers.....”*

SMS from a Mom whose child has an as yet undefined serious PID

- *“Good evening doctor. This is K’s mommy. I don’t understand the diagnosis. Is there something (they) are not telling me ?”*

SMS from the mother of an adolescent being investigated for unexplained bronchiectasis

- *“Thank you for helping, we appreciate it!!!! He was in so much pain, I did not know what to do anymore”*

SMS from the concerned wife of an adult patient being diagnosed with an autoinflammatory condition.

Many Questions About Diagnosis and Treatment

to prepare the
visit

SPECIFIC TO THE PID

- What else could cause these symptoms ????
- What is the specific type of primary immunodeficiency I have?
- How can I keep my child from getting sick?
- What reactions should I expect from the treatment?
- What treatments are recommended and available for this PI and why?
- Are there alternatives to this treatment approach?
- How long will treatment be required?
- How will I know if treatment is working?
- Will we be able to change treatment or product later , if not tolerated?
- Should my child/family get all the usual vaccinations still ?
- Does one need to stay away from some sports?

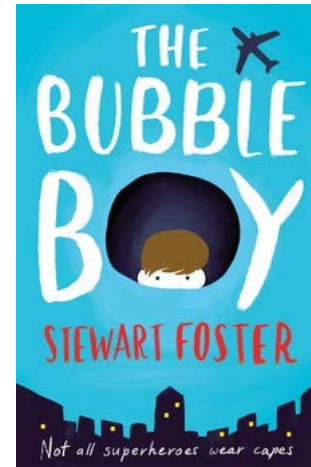
**And always
the worry :**

Is it SCID ????

**Will I, he she
die soon ?**

SCID is a very serious , but a rare PID .

The white blood cells called lymphocytes are very low or almost absent then, with onset in infancy usually



What is “IVIG” ?

Most common replacement for PID

- **Immunoglobulin (Ig) replacement therapy.** These are disease-fighting antibodies (proteins), given through a needle, by intravenous infusion in hospital/centre.
- These (passive) antibodies **only last a while** , so you/your child may need treatment **every 3 or 4 weeks**.
- There may be some side effects such as aching muscles or joints, headaches, or a low fever.
- **Subcutaneous Ig infusion** needs training but can be **given at home** into the deeper skin tissue and side effects are rare.

What treatment

Stem Cell Transplants



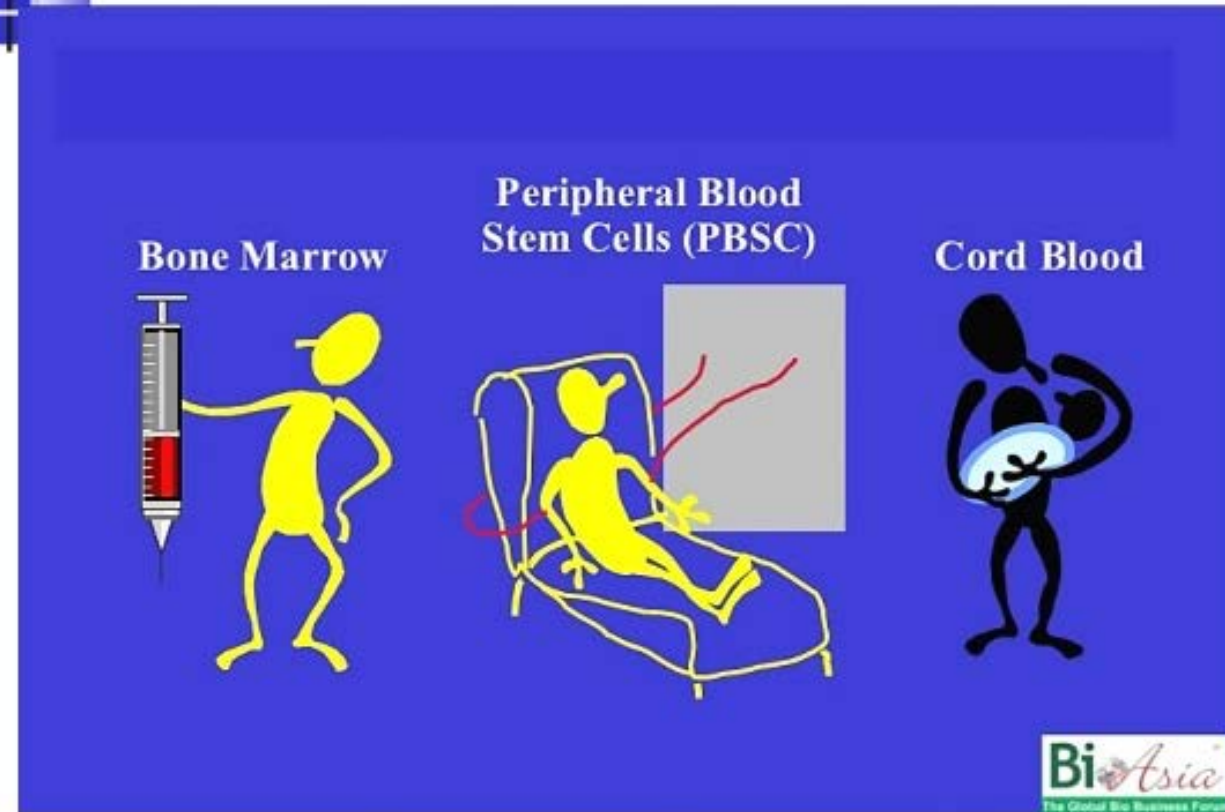
?

For severe PID like SCID

- **Stem Cell transplant.** For a very severe PID, your child may need a stem cell transplant urgently.
- Stem cells help make new blood cells. They come from bone marrow in the centre of bones.
- For a transplant, a donor supplies stem cells which have normal function. A donor needs to be the right match so that the body accepts the new cells.
- Close relatives, such as a brother or sister, are the best donors. Someone from the same racial or ethnic background may also be a good donor.

Stem cells for transplant do not always have to be a “Bone Marrow” donation. They are other sources too.

Sources of Hematopoietic Stem Cells

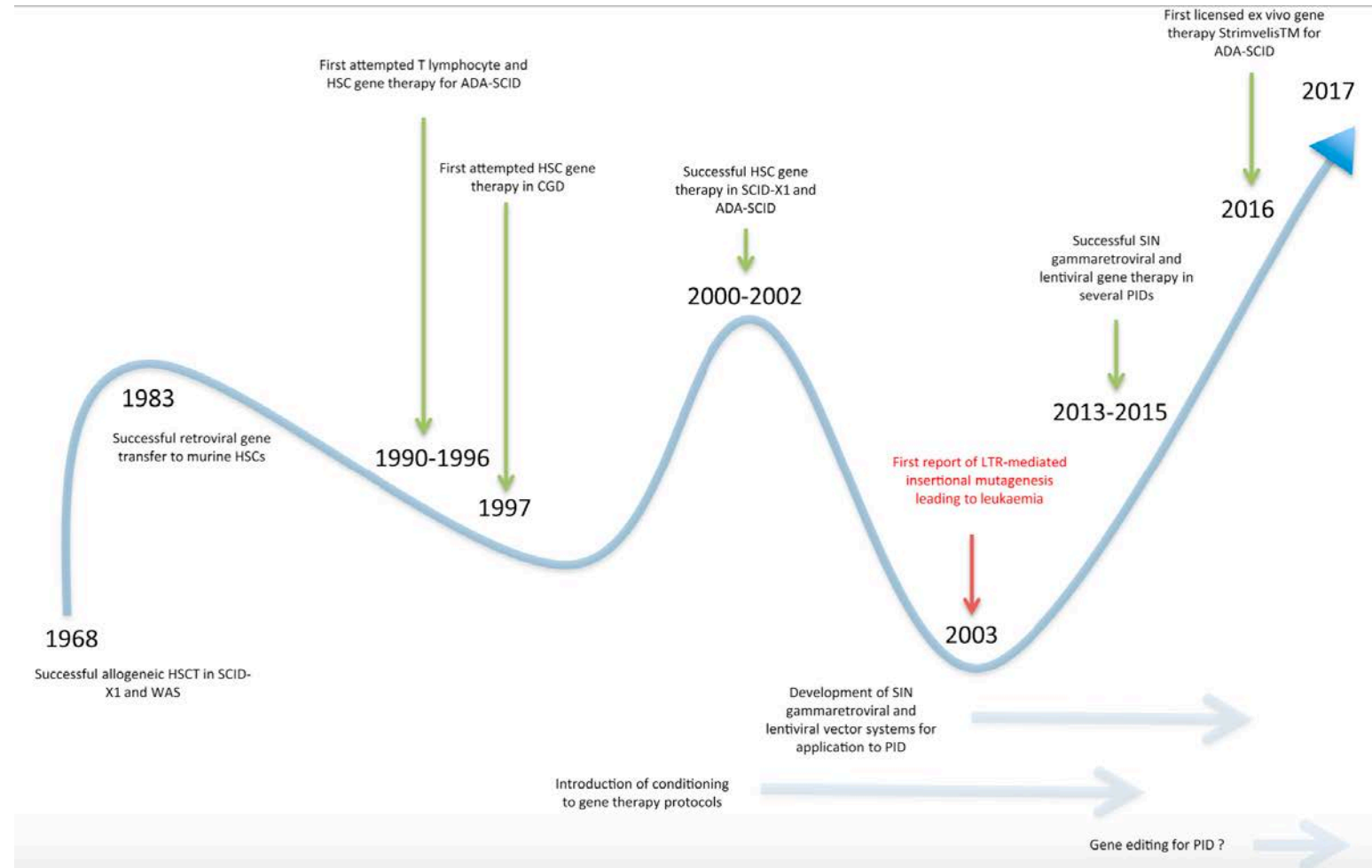


What about Gene Therapy ?

- Get a genetic diagnosis/counselling if possible to define the PID precisely, or ask if blood can be stored for future genetic work.
- It can affect other family members too who may then need screening
- The future holds promise for correction of many PID genes with minimal side effects



History of Gene Therapy



Molecular Therapy
Volume 25, Issue 5, 3 May 2017, Pages 1132-1141

Prepare further Questions About

Living With PID

- Who will be part of my healthcare team? Who will be my main point of contact?
- Whom should I call first if I get sick?
- Are there any restrictions to activity or diet? Is there a specific medical diet or are there nutritional guidelines you would recommend?
- Is there a known genetic component for this type of PID? If so, how will I know if other family members need to be tested?
- What are the long-term health consequences of my /our PID?
- Is there a PID support group I can join?
- Do I need to see any other specialists?
- Can I get help with insurance related inquiries, am I covered by state health ?
- Does my medical aid cover my/our condition (ICD10 code) as a benefit?

General Points

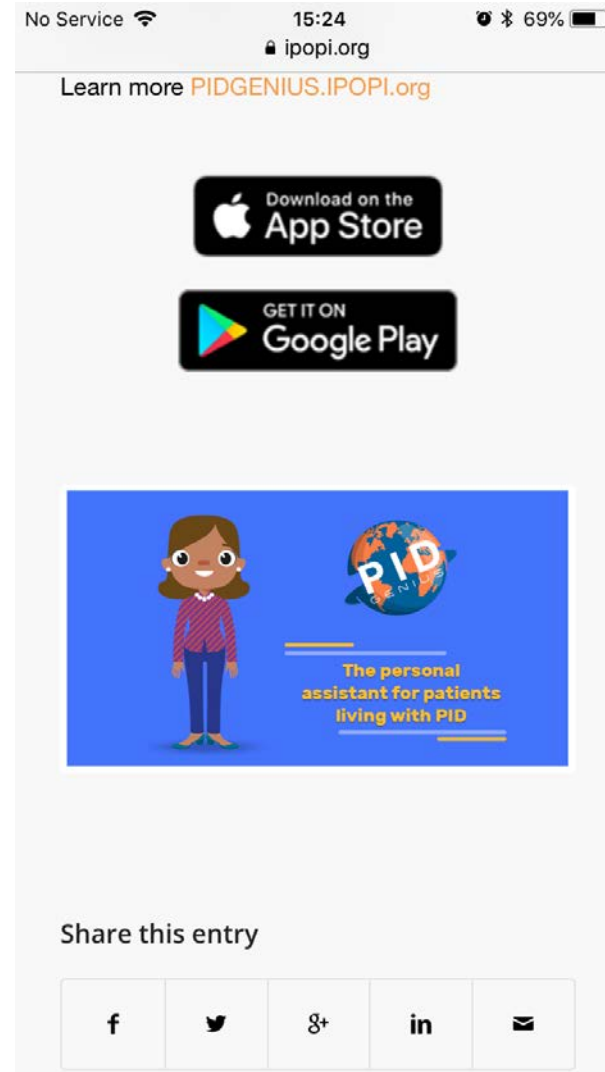
- Live vaccines are best avoided until assured by your specialist, but killed vaccines should not be avoided
- Careful of : Supplements, vitamins, immune boosters, homeopathy supplements etc etc without proven benefit but a lot of expense
- Antibiotic prophylaxis (low dose) may be useful or indicated
- Use only BOILED water if you have a PID
- Find and stay with one primary physician whom you trust to coordinate investigation and treatment referrals
- Find your support group !

Recommendations

- Start a FILE – Stay in CHARGE !
- Keep a dedicated diary – doctor's visits (names, speciality seen), infusions, X-rays, special/routine procedures



IPOPI Mobile App “PID GENIUS”



Many Sources



- IPOPI Website
 - JMF Website "Village"
 - Immune Deficiency Foundation: "Patient & Family Handbook."
 - PID UK
-
- Acknowledgement for some of the slide content to the IDF & SHIRE leaflet on "Questions"



THANK YOU