

Taking Charge of Your Care and Learning How Important Health-Related Quality of Life Is in Primary Immunodeficiencies (PI)

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Primary Immunodeficiencies

- Currently there are ~500 genetic disorders known as PIs¹
- Most of these PIs have variable clinical presentations including, recurrent or severe infections, and autoimmune or autoinflammatory complications such as:
 - Swollen lymph nodes or enlarged spleen
 - Gastrointestinal disorders
 - Low blood counts
 - Failure to thrive
 - Developmental delays





The Underlying Cause of Activated PI3K-Delta Syndrome (APDS)

- Inside every B and T cell in your immune system there is a protein known as PI3K delta²
- PI3K delta tells your B and T cells how and when to activate and mature, so that they can do their job²:
 - Having functional, mature B and T cells are important in detecting and killing viruses, bacteria, and other pathogens
- Patients with APDS have a hyperactive PI3K delta protein. This hyperactivity gives the B and T cells mixed messages about how to develop and function properly³:
 - This results in too many immature B and T cells which are not effective in fighting infections⁴
 - It can also cause the B and T cells to attack you, or accumulate in your lymph nodes leading to swollen lymph nodes and even swollen organs⁴
- One small protein (PI3K delta) can have big effects and can profoundly impact patients' health-related quality of life (HRQoL). Patients may experience recurrent infections which limit social, school and work activities and can cause symptoms such as fatigue⁴⁻⁶:
 - Fatigue, which is the feeling of being overly exhausted and/or having low energy, can interfere directly with one's ability to carry out routine daily activities

How You Feel Is an Important Symptom and Partnering with Your Healthcare Team Empowers You

- Several factors such as genes, childhood experiences, personal and working environment, relationships, lifestyle, diet, exercise, and sleep can affect how we function
- HRQoL can directly affect disease outcomes in PIs⁷:
 - This is critical because there are many factors which contribute to HRQoL, such as stress, sleep, and diet, that you might be able to modify⁵



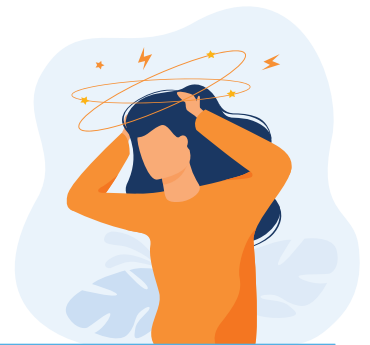
STRESS

Research has shown that stress can impact vaccine responses, and worsen viral and bacterial infections.⁸

- **Stress is less about what's happening and more about how we perceive what's happening⁹**
- **If we change how we perceive something, then we may be able to reduce how stressful it is⁹**

5 key-steps to change our perception include⁹:

- *Feeling in control (while you cannot control having a genetic disease, there are factors you can control)*
- *Feeling like the situation is predictable (establish routines to gain a sense of predictability)*
- *Feeling like things are improving (studies show that optimism can directly affect stress levels)*
- *Feeling socially connected (interacting with people who understand your challenges is powerful)*
- *Making sure you have an outlet (this can be a hobby, exercise, or the ability to discuss your frustrations with someone)*



FATIGUE

Fatigue can contribute to poor HRQoL, reduced work and/or school productivity and therefore can affect economic stability.⁵

- *Fatigue is an important symptom and can provide insights to you and your healthcare provider about what is going on inside of you, therefore reporting your fatigue is valuable. For example:*
 - » *Fatigue may indicate something called a “wear off effect” with your immunoglobulin replacement therapy (IRT)¹⁰*
 - > *IRT may be wearing off towards the end of the infusion cycle*
 - » *Fatigue can be linked to lower counts of immune cells and higher rates of non-infectious complications in patients with PIs (reduced digestive, kidney, liver, lung, and neurological function)¹¹*
- *If you are experiencing fatigue that does not improve with lifestyle modifications such as sleep or exercise, you may want to partner with your HCP to investigate if any of the above are happening to you*



SLEEP

Getting adequate amounts of sleep (7-10 hours per night depending on age) can help to improve.^{12,13}

- Brain function
- Hormone levels
- Immune function
- Overall metabolism



NUTRITION/MICROBIOME

PIs often present with gastrointestinal symptoms, which include infections, malnutrition, malabsorption, and changes in the microbiome.¹⁴

- **Patients with PIs have been described as having less variation in the types of bacteria that live in their gut, known as the microbiome¹⁵:**
 - Having increased variation in the microbiome is associated with having a more resilient gut⁴
 - The microbiome can affect your mood
- **Diet directly affects our microbiome, here are some key examples of foods that help support the gut microbiome:**
 - Fermented foods (eg, kimchi, sauerkraut, pickled foods)
 - Cultured foods (yogurt and cheese)
- **There are resources available to help you navigate an ideal diet for you to support your immune system and your microbiome:**
 - The Immune Deficiency Foundation provides key ways to eat healthy:
<https://primaryimmune.org/news/healthy-diet-exercise-key-general-care-pi>
 - The Harvard school of public health provides a yearly “healthy living” guide with detailed diet, exercise and lifestyle guidance:
<https://www.hsph.harvard.edu/nutritionsource/2022/01/06/healthy-living-guide-2021-2022/>



Support For People Affected by APDS

Finding support gives you a sense of community and access to tools, laws, and resources you did not even know were there for you.

- There are patient advocacy groups for patients with PIs such as the Immune deficiency foundation (IDF), that provide a range of supportive material and education:

<https://primaryimmune.org>

– IDF provides support on genetic testing: <https://primaryimmune.org/genetic-testing>

– IDF has a list of events that includes virtual support groups and meet-ups, as well as educational forums: https://community.primaryimmune.org/s/events?language=en_US

– IDF provides information regarding your rights as a patient with a PI in the work and school place:

<https://primaryimmune.org/primary-immunodeficiency-and-school>

<https://primaryimmune.org/pi-workplace>

– IDF also gives patients with PIs the opportunity to share their story:

<https://primaryimmune.org/share-your-story>



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