

International Coalition of Organizations
Supporting Endocrine Patients



Education and Truth

Global Awareness and Equity for
all Endocrine Patients



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<http://icosep.org>

INTRODUCTION: The World Health Organization recognizes that there are between 5,000 and 8,000 rare diseases. (1) Orphanet reports, that as of 2018, there were 6,000-7,000 identified rare diseases. 80% of those disorders are genetic in origin. (2) Globally, 350 million people suffer from a rare disease. (3) **Yet, less than 5% of all rare conditions avail treatment options.**

Patients and their families, patient organizations, medical providers and other members of ICOSEP realize the enormous challenges for this population of people. The desperate need for research and relevant treatment is significant for all rare medical issues and often overlooked.

PROBLEM: Why are new treatment options met with great resistance?

As a global coalition of endocrine related patient organizations, partnerships and networks, ICOSEP patient organization leaders have a shared belief that the resistance of health authorities to approve treatments for rare endocrine conditions is primarily twofold.

First Cause: Lack of proper understanding.

Scientists, medical providers, and others write literature for people with similar mindsets. For example: Medscape defines and describes Adult Growth Hormone Deficiency as follows: “GH deficiency in adults usually manifests as reduced physical performance and impaired psychological well-being. It results in alterations in the physiology of different systems of the body, manifesting as altered lipid metabolism, increased subcutaneous and visceral fat, decreased muscle mass, decreased bone density, low exercise performance, and reduced quality of life.

Adult GH deficiency can be a transition from childhood-onset GH deficiency or it can be acquired during adulthood. The majority of cases are caused by pituitary tumors or by their treatment with surgery, radiation therapy, or both. Traumatic brain injury is another important cause.

The goals of GH therapy in adults are to improve conditioning, strength, body composition, and quality of life, as well as reduce the burden of associated medical conditions such as cardiovascular disease and decreased bone mineral density. (4)

How much of this information did you truly grasp about this endocrine condition? Probably very little!

What this large explanation means to real patients is: If you are GHD as an adult, your body cannot make enough hormone to maintain your heart, lung and other vital muscles for your age. You cannot heal properly and your body will age and deteriorate much more rapidly than it should as you age. Your quality of life as a 60-year-old could be that of a much older person!

Second Cause: Calculations that “Rare” is Less Important because the number of patients is smaller. (5)

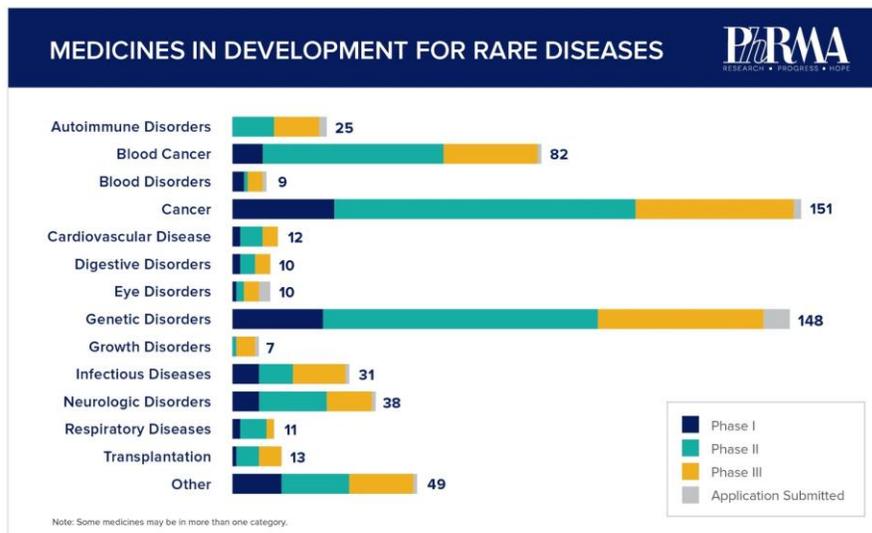
Throughout the world, a majority of people are not aware that hormones (endocrinology) impact every living cell and system in our bodies. The impact of the endocrine system may not have the immediate effects as going without oxygen would, but the damage to a body if the endocrine system is failing, is such as severe.

Endocrinology is a less known medical specialty which is under recognized for its importance. Each and every day we are all impacted by our endocrine system in ways we never realized before. For example, anxiety is rarely seen as being an endocrine issue. If diagnosed properly and treated for true endocrine problems, patients may be able to prevent serious emotional problems. (6) Another example is adults

who have hypertension (high blood pressure), which is a risk factor for heart disease may in truth, have an endocrine basis for their medical issue. It is well documented that as many as 10% of the people who have hypertension may have this condition because their body is producing too much aldosterone (a hormone produced in the adrenal glands). (7) But how many people understand that the endocrine system needs to be considered...or do they simply begin blood pressure medication without thought?

The lack of identification pertaining to endocrine conditions is so large and ever changing that no medical database nor research entity has ever successfully compiled a registry of all the diseases in this area. The only known global resource list is available from ICOSEP <https://icosep.org/resources/>.

In the graph which follows titled *Medicines in Development for Rare Diseases*, the category which encompasses endocrine issues is named growth disorders. The number of medicines in development for this patient population is comparatively low. Even the autoimmune category is highly impacted by the endocrine system. (8) Also the category of Neurologic Disorders is closely related to endocrine issues as “endocrine dysfunctions may lead to various neurologic manifestations “. (9)



It is ICOSEP’s position that available treatments for endocrinology issues are highly overlooked and unfortunately dismissed due to a lack of educated data. Quick judgements or approval for temporary treatments to fix symptoms rather than the root cause of the symptoms are readily accepted and patients suffer. Up to date treatments such as weekly therapies compared to daily therapies are met with skepticism despite the equality in costs and improved quality of therapy results.

Treatment adherence is a complex, multi-factorial process that is notoriously difficult to quantify, due to its complexity and the inherent bias of monitored administration. Studies focusing on therapy-related interventions consistently point to dose frequency, treatment complexity, and reduced patient compliance with therapy as key barriers to adherence (World Health Organization,

2003, Haverkamp et al, 2008; Cutfield et al, 2011; Fisher and Acerini, 2013). Increasingly, there is a need to develop, ‘Minimally Disruptive Medicines’ to address the demands placed on patients managing chronic disease. (May et al, 2014; Abu Dabrh et al; 2015; Leppin et al, 2015; Serrano et al, 2017). By reducing treatment frequency, endocrinology treatments provide a treatment option for patients that may increase their compliance with therapy, and thus improve treatment outcomes. There are

mountains of scientific data which prove that patients (of all sorts and conditions) prefer once a week treatments as compared to daily regimens. Patients are much more likely to follow the physician's orders without falter, thus reducing potential adverse effects of missing treatments and reducing potentially costly future complications.

It is also well documented that less intrusive weekly therapy is equally effective or higher than the daily treatments. One study reported "Most patients believed that once-weekly dosing was more convenient than once-daily dosing (89.0% vs 7.7%; $P < 0.001$) and would allow them to achieve better long-term compliance (87.5% vs 8.5%; $P < 0.001$)." (10)

As a patient coalition, members of ICOSEP do not believe that authorities responsible for allowing new procedures or treatments are politically motivated nor malicious as these medical problems may impact their families too. However, we do firmly believe that honest education for the health ministries and medical professionals is essential. Only after review of true facts, can decisions be made which will have the greatest impact on the patients' lives and hopefully reduce long term medical complications.

This position paper is agreed upon and signed by the following organizations. (More to be added.)

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Resources:

1. https://www.orpha.net/consor/cgi-bin/Education_AboutRareDiseases.php?lng=EN
2. <https://innovation.org/about-us/commitment/research-discovery/rare-disease-numbers>
3. <https://globalgenes.org/rare-facts/>
4. <https://emedicine.medscape.com/article/120767-overview>

5. <https://www.multiplesystematrophy.org/uncategorized/rare-disease-day-2018-importance-funding-research/>
6. <http://www.drryanhall.com/anxiety.htm>
7. http://www.socalendocrine.com/Endocrine_Diseases.html
8. <https://www.ncbi.nlm.nih.gov/pubmed/3512901>
9. <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC4316409/>
10. <https://www.ncbi.nlm.nih.gov/pubmed/12501880>