
A Picture Paints a Thousand Words, But with Rare Diseases There Are a Million More: Will You Help the Global Cushing's Patient Community Educate the Public About Cushing's Syndrome?

*Cushing's Syndrome is the most difficult endocrine disease to diagnose and treat and involves a tumor or other source causing the adrenal glands to make too much **cortisol**, the "fight or flight" and stress management hormone. Cushing's lacks definitive markers, and though it has over three dozen potential symptoms, they are all non-specific and many are common today such as high blood pressure, diabetes, weight gain, insomnia, cognitive/memory problems, anxiety, anger, and depression.*

LILBURN, Georgia, February 29, 2024: Cushing's Support and Research Foundation (CSRF), a global key opinion leader in patient advocacy and quality of life research, invites interested media to connect for a deeper exploration of the patient point of view on the complexity of this diagnosis.

There is an antiquated saying in the medical community to "look for horses when you hear hoofbeats, not zebras", and this thinking contributes to diagnosis delays for Cushing's patients that average five or more years. Some forms of Cushing's are rare, and there is ongoing research looking at all forms of hypercortisolism. A recent Phase 4 clinical trial enrolled participants with difficult-to-control type 2 diabetes, tested their cortisol, and found that 24% had Cushing's Syndrome. [1] What could this mean for society – how many people are walking around with difficult-to-control, non-specific "metabolic issues" that are not setting off a cortisol testing alarm?

It's not possible to fully understand what Cushing's feels like unless you have it. It is full-system; we have cortisol receptors in nearly every part of our bodies. The longer we have Cushing's, the worse our long-term health gets; the journey does not end with resolution of the excessive cortisol. Diagnostic tests are inexpensive and at least 94% accurate [2]. So why is this not getting picked up faster by primary care doctors, gynecologists? **Why did it take merciless bullying about her appearance for Amy Schumer to be diagnosed?** The entire global network of patients came alive when she announced her diagnosis, even though she called hers "the one that goes away".

CSRF is a U.S. 501c3 non-profit support organization formed in 1995 and currently run by patients with an exceptional Medical Advisory Board [3]. CSRF's mission is to connect with patients and their caregivers for education and support and to engage the medical and research communities to supply new quality of life data to change outcomes. CSRF officially broke ground on the first ever global patient registry project in January 2025 with a special funding award from the Critical Path Institute, the National Organization for Rare Disorders (NORD), and the FDA [4].

References:

- [1] Frias JP, Auchus RJ, Bancos I, Blonde L, Busch RS, Buse JB, DeFronzo RA, Findling JW, Fonseca V, Hamidi O, Handelsman Y, Pratley RE, Rosenstock J, Tudor JC, Moraitis AG, Einhorn D. **FRI183 Phase 4 Study Of Hypercortisolism In Patients With Difficult-to-Control Type 2 Diabetes Despite Receiving Standard-of-Care Therapies: Prevalence And Treatment With Mifepristone.** Journal of the Endocrine Society, Volume 7, Issue Supplement_1, October-November 2023, bvad114.178, <https://doi.org/10.1210/jendso/bvad114.178>
- [2] Galm BP, Qiao N, Klibanski A, Biller BMK, Tritos NA. **Accuracy of Laboratory Tests for the Diagnosis of Cushing Syndrome.** J Clin Endocrinol Metab. 2020 Jun 1;105(6):dgaa105. doi: 10.1210/clinem/dgaa105. PMID: 32133504.
- [3] CSRF Medical Advisory Board. <https://csrf.net/about-csrf/medical-advisory-board/>. Accessed February 28, 2024.
- [4] Rare Disease Cures Accelerator. <https://rarediseases.org/rdca-dap/>. Accessed February 28, 2024.

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