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After all, Porphyria exists in Portugal! A three-year study

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Introduction: Porphyrias are a group of eight rare inherited disorders, each caused by a defect in a specific enzymatic step of heme biosynthesis. These disorders are multisystemic, with variable symptoms, and represent a major burden for patients and families, with disabling chronic symptoms scattered with life-threatening acute attacks. There are two main clinical types of porphyria: acute porphyria and cutaneous porphyria. Acute porphyrias are often misdiagnosed because of their diverse clinical manifestations, which can mimic other diseases (1).

Methods: Porphyrin precursor accumulation patterns and total urine porphyrins (TUP) are the first-line laboratory tests. The determination of porphyrin profiles in biological samples and the plasmatic emission fluorescence peak are the second-line tests. The NGS porphyria panel is the third-line test.

Results/Case report: In Portugal, our unit (URN-INSA, Porto), also an associate member of IpNet (International Porphyria Network), is currently considered the reference laboratory for the biochemical and molecular characterization of porphyria. Since 2022, a cohort of 139 patients has been screened for porphyria. The development of acute and cutaneous diagnostic algorithms has resulted in 34 porphyrias: 5 cases of Acute Intermittent Porphyria (AIP), 1 Variegata Porphyria (VP), 2 Hereditary Coproporphyria (HCP), 23 Porphyria Cutanea Tarda (PCT) and 3 Erythropoietic Protoporphyria (EPP).

Conclusion: Even so, these figures are lower in comparison to other similar countries, as we should have a higher prevalence. This diagnosis was not available in our country, which is now possible at URN-INSA (2). From this work, we have concluded that the articulation between the clinician and the laboratory is crucial to choosing the right biochemical test to achieve the correct diagnosis and complete characterization of the disease. Porphyria exists; we just have to look for it (3)!

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