Editorial

Rediscovering the essential: the silent challenge of Familial hypercholesterolemia in Latin America.

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ABSTRACT

Familial hypercholesterolemia (FH) remains widely underdiagnosed in Latin America, despite its high prevalence and strong association with premature cardiovascular disease. The study conducted in a subsidized population in Bucaramanga, Colombia, using the Dutch Lipid Clinic Network (DLCN) criteria, reveals the gap between clinical suspicion, genetic confirmation, and therapeutic control. Despite high adherence, only a third of patients achieved LDL-C goals, underscoring the complexity of the phenotype and the need for structured strategies. This editorial discusses the regional implications of these findings and emphasizes the need to strengthen screening, expand access to genetic testing and promote the development of lipid clinics. FH remains an unmet need in the region, and early detection represents a concrete opportunity to change the natural history of the disease.

Familial hypercholesterolemia (FH) is one of the most significant genetic disorders in cardiovascular prevention. Its impact does not spring only from its prevalence, higher than what was assumed for years, but from the lethal combination between a delayed diagnosis, a prolonged exposition to extreme levels of LDL-C and a natural history marked by premature CAD. In a continent where atherosclerotic disease is still the main cause of death, FH is a clinical challenge that remains silent, to a good extent because it is not sought with the necessary systematicity.[1,2,3]

The study made in Bucaramanga provides several layers of reflection. First, it shows that the Dutch Lipid Clinic Network (DLCN) criteria, widely validated as a clinical tool, are still an efficient way to identify patients in risk, even in health care systems with limited resources.[2,3,4,5,6,7,8] The evaluated cohort is representative of a frequent Latin American reality: elder patients, with a high load of co-morbidities, cared for in contexts where access to specialized follow-up is irregular. For only twenty people to meet the suspicion criteria does not imply a low prevalence; on the contrary, it reveals the structural problem: most are never even evaluated with an approach toward FH.

Another aspect deserving attention is the therapeutic gap. Adherence, close to 90%, suggests an appropriate commitment to treatment. However, only 30% reached LDL-C goals. This imbalance, observed also in international registries, exposes to converging realities: diagnostic inertia, patients arriving late, and therapeutic inadequacy before the genetic phenotype.[3,4,9,10,11,12,13] Statins, even in high doses, are usually insufficient in patients with severe LDLR mutations. And without an appropriate diagnosis, stronger therapy combinations are hardly proposed.

The genetic component of the study enriches the clinical view. Identifying pathogenic variants in LDLR, along with APOE and PCSK9 mutations, confirms the value of genetics as a supplementary tool. Also, it displays a reality known by those working in the region: global databases do not reflect properly the Latin American genetic diversity, essentially associated to the high cost of genetic testing or the non-availability of the technique in said countries, reducing the identification of the variables known to be pathological, besides a significant number of variants of uncertain meaning, which may explain the behavior of this pathology in the region.[1,2,14] This lack of representation leads to mutations being interpreted without a local population reference, limiting diagnostic accuracy. In spite of this, the information provided by molecular analysis is determinant to adjust therapy strategies and to start a family research, a crucial element in hereditary diseases.

The social dimension of the study is not to be overlooked either. Most patients belonged to low socioeconomic strata, a pattern that keeps repeating in Latin America. Financial

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vulnerability limits access to tests, consultations and medications, and increases the chance for a diagnosis to be delayed for decades. The result is predictable: premature CAD, disabilities and death in ages where an active life is to be expected. FH, that could be detected in adolescence, is still diagnosed after the first coronary event, when damage is already significant.[11]

Faced with this scenario, primary care emerges as an indispensable protagonist. It is clinicians, internists and clinical cardiologists who should suspect FH in the presence of persistently elevated LDL-C, a family history with early infarctions or the presence of xanthomas.[10,12] Without the participation of the first level, no research system should prosper.

Likewise, the study points out the chance and the need to strengthen the lipid clinics in the region. These spaces allow to provide specialized care, adjust high-intensity treatments, coordinate cascade screening and perform continuing education. Where they exist, detection increases and results change. Where they don't, FH remains invisible.

The Bucaramanga experience shows that even with budgetary restrictions, directed genetic testing can be implemented, with a real clinical impact. It is an encouraging message for the region: no abundance of resources is required, but clarity in priorities.

Finally, this study is a reminder. FH is preventable, detectable and treatable. It is not a rare disease. It is not an incidental finding. It is a problem of public health. Underdiagnosing it is not due to lack of evidence, but to a lack of implementation. And it is precisely there where Latin American cardiology is called to lead a change.

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