Lipoedema: A genetic disease to know better

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The discovery of the first gene (AKR1C1) responsible, if mutated, of Lipedema (from the greek word ‘oedema’ which means swelling and no ‘accumulation of fluids’), has definitively removed the doubts on the genesis of the pathology of 'pure Lipedema', that is, of the clinical picture that does not present overlaps with obesity or other secondary comorbidities to the disease. The BMI in most of these pictures is normal and it is also possible to observe cases of women at the limits of anorexia for fear of increasing the volume of the affected areas and the pain that accompanies this.

Speaking of pain, there are those who argue that there is a mental disorder at the base. In our experience, pain is an integral part of the initial clinical picture in most cases. Mental disorder is a consequence of clinical distress. The same first enzyme discovered to be responsible for Lipedema, if mutated, is unable to catabolize Pregnanolone, a neurosteroid that has an analgesic effect by increasing GABAA currents. It is no coincidence that the three mutated familial cases described in the article illustrating the discovery did not present pain.

It is necessary to investigate all these aspects in order to better define the pathogenesis of the disease, not linked to behavioral disturbances on the part of the patients but to familiarity with genetic transmissivity.