GENETICS AND GENODERMATOSES

EHLERS-DANLOS SYNDROME, A SINGLE CENTRE EXPERIENCE

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BACKGROUND: Ehlers-Danlos syndrome (EDS) is a group of congenital diseases with collagen alterations. Cutaneous fragility, hyperextensibility and joint hypermobility are the characteristic triad. In 2017 a new classification with 13 entities was done based on the clinical and genetic characteristics. The aim of this transversal, descriptive study is to present experience in our service with EDS.

OBSERVATION: 12 patients fulfill clinical and/or genetic criteria for EDS; 5 men and 7 women. Mean age was 25 years-old. In 4 of them genetic tests were done: one presented a PLOD1 mutation, another was negative, one is still waiting for genetic results and in the least, further genes have been asked. In two patients, a definitive diagnosis was done for PLOD1 and hypermobile EDS (hEDS) respectively. The more common no-dermatological findings were dysautonomia and luxation, present in 66% of patients, gastrointestinal dysfunction (58%) and column alterations, fractures and/or muscle hypotonia (50%). Dermatological more common findings were pseudo-tumors, atrophic scar (58%) and skin hyperextensibility (50%). In one patient, a spontaneous femoral rupture took place and in another a concomitant diagnosis of hyperhomocysteinemia was done.

KEY MESSAGE: A high clinical variability and overlapping is present between EDS types. That is why differential diagnosis between EDS types and with other hereditary connective tissue diseases like cutis laxa, Marfan syndrome, Loeys-Dietz syndrome and others, represents a challenge. For this reason, genetic test is necessary for a correct diagnosis, which will determine the correct management and multidisciplinary follow up of the patient.