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Claude Hamonet

University of Paris-Est-Créteil, France

Ehlers-Danlos and neurological diagnosis

Aim: Ehlers-Danlos is a hereditary disease, multisystemic and frequent of the collagen tissue, which excludes the nervous system. However, a deceitful symptomatology of this disease, poorly known and described will often lead the practitioner towards a neurological or psychiatric pathology with iatrogenic risks.

Method: Our experience is based on a cohort of 5,600 patients, women (80%), men and children with an important alteration of their collagen tissue, including meningeal (with a fragilization of the brain) and a proprioceptive disorder with hyper sensoriality at the origin of their symptoms. 5 of the 9 following clinical signs allow for a precise diagnosis at 99.6% without reliable genetic testing, rebellious and widespread chronic pain, chronic fatigue, difficult motor control with frequent clumsiness, joint instability, skin fragile and thin, hypermobility joints, ecchymosis, gastroesophageal reflux and hyperacusis. We have spotted erroneous neurological diagnoses with our patients.

Result: The orientation towards a mental illness is the most frequent case, due to the multiplicity and variability of the symptoms but also due to cognitive difficulties (memory, attention, concentration), affectivity disorders (anxiety) associated with a physical and intellectual hyper activity. The motor difficulties (bladder and sphincter, visual with diplopia, deglutition) often lead toward a diagnosis of multiple sclerosis. Spinal and limb pain, frequent in EDS, leads to a diagnosis of compression of roots, the brachial plexus or the nerves (median, ulnar). Frequent dystonia (66%) brings to mean Parkinson disease or epilepsy. The latter is also evoked due to of loss of consciousness as a result of dysautonomia. The pseudoparalysis (proprioception) is confused with a medullar compression, a polyradiculoneuritis or a stroke. The muscles signs are often confused for muscular dystrophy or myasthenia.

Conclusion: Due to its high frequency (incidence: 2% in France, 3% in Belgium), its painful symptomatology and deceitful motor, the Ehlers-Danlos disease should largely be known to neurologists, so that it may be quickly identified.

Discussion: Erroneous psychiatric or neurologic diagnoses are frequent with EDS patients and this disease should be systematically evoked in the discussion of these neurologic diagnosis. This will avoid therapeutic accidents (hemorrhages due to platelets anti-aggregating treatments) and a worsening of EDS for which different treatments exist (proprioceptive garments and orthosis, oxygen therapy). The current importance placed on the psychosomatic phenomena should be minimized and replaced with a greater focus on listening to the patient.

Biography

Claude Hamonet is currently Emeritus Professor, University Paris-East-Creteil (UPEC), and worked as Dean of Faculty-UFR Communication and Social inclusion University Paris12 (1985-1992). Research expert for Found of Health researches and Ministry of Universities in Quebec. Director of French reference centre associated with national reference centre of EDS (2005-2007). Consulting Physician, Ehlers-Danlos disease, in ELLAsanté medical center Paris

pr.hamonet@wanadoo.fr