**CO44-003–EN**

**Reference center spina bifida**


*CHU Rennes, France*

**Keywords:** Spina bifida; Spinal dysraphism; Myelomeningocele; Meningocele; Tethered spinal cord

**Patients and methods:** A multidisciplinary team has set up a certified National Reference center for rare diseases. Specialties within the Center are: Physical medicine and rehabilitation, proctology, urology, neurosurgery, orthopaedic surgery, genetics, obstetrics and gynecology, sexology, dermatology, plastic surgery. A national network has been established based on the recognition of nine regional expert centers. Relationships with users are made through patients associations, especially with ASBH.

**Results:** Two hundred and five people with spina bifida (197 adults and eight children) consulted the Center of reference. They were then referred to the nearest regional center. The management was made in connection with the community medical and social services. The main reasons for consulting were: sphincter dysfunction (urinary and rectal), walking impairment, pain, global demand for rehabilitation therapy where each individual was his/her own control.

**Discussion:** The syndrome is very often confused with fibromyalgia, sclerosis, axial rheumatism, asthma, Crohn’s disease, hypothyroidism, and psychopathology. Very rare forms with a vascular, intestinal, obstetrical important risk have been described but the distinction remains unclear despite the identification of COL3A1 in vascular EDS. The therapeutic contribution of garments is confirmed.

**Further reading**


**CO44-005–EN**

**Management of patients in the Angers ALS Center**

G. Nicolas, M.-L. Le Peillet, B. Rivron

Département de neurologie, centre SLA d’Angers, 4, rue Larrey, 49933 cedex 9 Angers, France

*Corresponding author.

**Keywords:** ALS Center; Management of patient; Occupational therapist

**Results:** – Described by Edvard Lauritz Ehlers (1900), then by Alexandre Danlos (1908) this debilitating genetic connective tissue disease is artificially designated by two signs: hypermobility and hyperlaxity. The diagnosis is purely clinical, based on the association of pain, fatigue, impaired proprioception, fragile skin hypermobility, bleeding, constipation, gastric reflux, dyspnea, respiratory “blockage”. Other events: ENT, ophthalmology, cardiovascular, obstetrical, bladder, spine, thermal, hypnic, memory, attentional disorders, are also observed.

**Discussion:** – Compressive garments, braces, TENS, “Percussionnaire”, oxygen, balance therapy.

**Further reading**


**CO44-004–EN**

**Ehlers-Danlos Syndrome (EDS), a new clinical description, efficiency of physical medicine and rehabilitation. Six hundred individuals studied**

C. Hamonet

Hôpital Raymond-Poincaré, Hôtel-Dieu de Paris, médecine physique et de réadaptation, 1, place du Parvis Notre-Dame, 75181 cedex 04 Paris, France

**Keywords:** Rare genetic disease; Ehlers-Danlos Syndrome; Hypermobility; Physical medicine and rehabilitation; Orthosis; Haemorrhagic syndrome

**Objectives:** To redefine the symptomatology of Ehlers-Danlos syndrome and help better identify it. Propose and develop treatments mainly in physical medicine and organize rehabilitation.

**Patients:** A study of 600 patients with active file followed by physical medicine and rehabilitation units, examined by the same physician, according to a standard analytical and quantitative evaluation. Production of a database and use of Excel software. Evaluation of rehabilitation therapy where each individual was his/her own control.

**Results:** – By examining 600 individuals with EDS, a new clinical description has been made, which defines EDS as a progressive, chronic, life-threatening, rare genetic disorder. The clinical picture is multifaceted and includes major life-threatening manifestations and minor signs which are very common in physical medicine and rehabilitation (AMR) centers. The Ehlers-Danlos Syndrome is a classical disease of connective tissue, involving many organ systems. The symptoms are not specific, and cannot be attributed to other causes.

**Discussion:** – The diagnostic criteria are based on the presence of at least 4 of the 9 criteria described by Zlotogora et al. (1998): hyperlaxity, hypermobility, hyperextensibility of joints, hyperelasticity of skin, hyperpigmentation of scars, telangiectasia, vascular problems, hyperextensibility of the skin, extravasation of blood or body fluids. These criteria are not specific and can be found in other connective tissue disorders.

**Further reading**

Corresponding author.

**Keywords:** Reference center; Child; Limb malformations of children; Arthrogryposis

**The national reference center for rare diseases regarding limb malformations of children and arthrogryposis**

F. Guillou, D. Pilliard

*Service de rééducation orthopédique de l’enfant, hôpital national de Saint-Maurice, 14, rue du Val d’Osne, 94415 Saint-Maurice cedex, France*

*Corresponding author.

**Keywords:** Reference center; Child; Limb malformations of children; Arthrogryposis

Since 1968, the orthopedic rehabilitation service for children at Saint-Maurice’s hospital treats children with limb malformations. In 2007, this department was certified as a reference center for rare diseases with expertise in congenital limb malformations such as total or partial agenesis of one or several limbs, bone misalignment caused by synostosis or asymmetrical malformations of one or several limbs. They also deal with ampu-