Reference center spina bifida

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

Objectives.– In France, approximately 16,000 people are diagnosed with spina bifida and related disabilities. The functional impairment is multiple and complex in spina bifida and is life-threatening. The Center was created to improve the coordination between specialists and to allow a smooth transition and continuity of care from childhood to adult age both on a locally and on a national base. Other aims are the promotion of education and research coordination.

Patients and methods.– A multidisciplinary team has set up a certified National Reference center for rare diseases. Specialities 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 (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 comprehensive care and information on prognosis, a query on sexuality. A satisfaction survey of patients showed an overall rating of 8.6/10. Patient records have been computerised to be shared between the different centers of the network and to feed a data bank; two main research projects have been jointly promoted at national level by the center of reference in conjunction with the regional centers. A process is underway on the definition of clinical guidelines.

Conclusion.– The action of the National Reference Center in conjunction with the Regional Experts Centers has improved care of spina bifida patients but further efforts will be required to reach the most dependent severely affected patients. The national and European networks have to be strengthened.

Further reading

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

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Keywords: Rare genetic disease; Ehlers-Danlos Syndrome; Hypermobility; Physical medicine and rehabilitation; Orthesis; 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.

(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 "blocage". Other events: ENT, ophthalmology, cardiovascular, obstetrical, bladder, spine, thermal, hypnic, memory, attentional disorders, are also observed.

Treatments.– Compressive garments, braces, TENS, "Percussionnaire", oxygen, balneotherapy.

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

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Management of patients in the Angers ALS Center

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Keywords: ALS Center; Management of patient; Occupational therapist

Founded by Professor Jean Emile in 1991, the ALS Center of Angers was officially acknowledged in 2003 along with 17 other centers during the "ALS-cystic fibrosis plan". In 2010, after a positive assessment by the French health authorities, the ALS Centers joined the "rare disorders plan", with the appointment of two referral centers (Paris and Marseille-Nice) and 15 "centre de competence" with funding. Organized as a multidisciplinary consultation, the ALS Center of Angers is in charge of more than 200 patients evaluated every 3 months. The ALS Center helps patients cope with disability and the announcement of this serious disease and its consequences. The occupational therapist plays a central role for patients facing an often rapidly evolving deficit, while anticipating the possible future disabilities. The rehabilitation specialist is occasionally requested for problems beyond the expertise of occupational therapists or other professionals in the ALS Center.

Monitoring of patients in ALS centers involves active collaboration with rehabilitation centers, local hospitals and other health professionals.

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

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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 agenesia of one or several limb segments, bone misalignment caused by synostosis or asymmetrical malformations of one or several limbs. They also deal with ampu-