CO44-003–EN
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 the national level by the center of reference in conjunction with the regional centers.

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


CO44-004–EN
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; 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 – 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 and hypermobility, bleeding, constipation, gastric reflux, dyspnea, respiratory “blockage”. 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

CO44-005–EN
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 compétence” 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.


CO44-006–EN
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 agenesis of one or several limb segments, bone misalignment caused by synostosis or asymmetrical malformations of one or several limbs. They also deal with ampu-
Development, activities and orientations of multidisciplinary consultations for neuromuscular diseases of the Nantes-Angers Reference center for rare diseases


Abstract:
Since the opening of the Nantes-Angers reference center for rare diseases in 2004, it has been able to manage these medical health procedures in association with other specialized centers (through meetings, shared information, censuses, medical protocols, research programs and joint publications). It may support other medical teams such as various prenatal diagnosis centers, surgical departments, medical physical and rehabilitation teams or general medicine. Outcome since its certification, we have recorded an increase in the patient workload. Numbers of consultations in 2007: 524–of which 33 antenatal consultations.

Number of consultations in 2010: 1011–57 of which antenatal consultations.

Certification has established a future for this unknown medical speciality and we can foresee breaking new grounds, especially in the area of research and in networking with fellow colleagues.

Keywords: Reference center; Neuromuscular diseases

Special ways of managing with neurodegenerative disease at home as seen by a mobile team of the Aquitaine serious disability network, HLA33

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Abstract:
Patients suffering from major neurodegenerative diseases living at home require multidisciplinary management and an adapted environment. By its expertise, a network can provide means and tools, and facilitate progress and implementation of care. It provides the necessary coordination between the professionals concerned.

Keywords: Network; Neurodegenerative pathologies; Living at home

Dependent patients discharged home from PRM departments: Relevant indicators

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Abstract:
Taking into account the complexity of the situation and the progressive nature of neuromotor and psychosocial disorders, this intervention network is an asset aiming to improve care, organise referral, facilitate coordination, offer educational and occupational training possibilities with the goal of improving the quality of life in the home environment.

Keywords: Hospital discharge; Dependence; Indicators; Coordinated care

Network; Neurodegenerative pathologies; Living at home

Introduction.– Patients suffering from major neurodegenerative diseases living at home require multidisciplinary management and an adapted environment. By its expertise, a network can provide means and tools, and facilitate progress and implementation of care. It provides the necessary coordination between the professionals concerned.

Keywords: Network; Neurodegenerative pathologies; Living at home

Objectives.– To present the problems most often met with by these professionals and the ways used by the HLA33 network.

Methods.– Starting from an example, a retrospective analysis of the medical records of patients suffering from neurodegenerative diseases due to parkinsonian syndrome or hereditary disease, compared with the entire group of patients followed by the network.

Results.– In 2010, the neurodegenerative diseases previously mentioned represent the third cause of disability of the patients in our network. From October 2004 to May 2011, out of 67 persons followed for these diseases (that is 10% of the patients in our network), there were 37 women and 30 men, on the average older than the general population of our network (55 years old).

Most of them were referred by a physician (neurologist, specialist in physical medicine or general practitioner). The patients were rarely referred directly after hospital discharge (16%) and most often lived in the Bordeaux area (73%).

The members of the staff were always called upon: the psychologist most often to assist the healthcare professional or the family caregiver, the occupational therapists for technical assistance and adaptation of home (renewed request as the disease progressed), the social worker for the needs concerning the human aids and financing. On average, work with these patients was longer than it was for the overall group in our files (18 months).

Seven patients died while they were followed by our network.

Discussion and conclusion.– Taking into account the complexity of the situation and the progressive nature of neuromotor and psychosocial disorders, this intervention network is an asset aiming to improve care, organise referral, facilitate coordination, offer educational and occupational training possibilities with the goal of improving the quality of life in the home environment.

Keywords: Hospital discharge; Dependence; Indicators; Coordinated care

Objectives.– To evaluate, based on existing recommendations, the criteria of a satisfactory organization when discharging to home a patient who remains dependent. It also aims at establishing simple and measurable indicators of the coordination between hospital and ambulatory care.

Patients and methods.– All the general practitioners of Maine et Loire (French administrative district) received a questionnaire asking them to rank the three main criteria (from the most to the least important) out of a choice of 14 [1,2,3] and offering them the possibility to add commentaries. We analyzed for each item the average, the standard deviation and median and highlighted the most important ones through an analysis of the relative frequency distribution.