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Chiari and Syringomyelia Consortium: a model of multidisciplinary and sharing path for Rare Diseases

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Abstract Piemonte and Valle d'Aosta Interregional Network for Rare Diseases (RD) is a model of "diffuse" network; it involves all the health system specialists devoted to the diagnosis, the therapy and the follow-up of a RD. The Consortium is a multidisciplinary team operating throughout the Region composed of volunteer Physicians and Biologists that promotes periodical meetings to develop shared protocols. In 2008 the Specialist-Technical Committee for the Interregional RD Network approved the set up of the "Chiari and Syringomyelia Consortium" (CSC) with two specific objectives: to identify the shared diagnostic criteria and to develop the interdisciplinary diagnostic-therapeutic-assistance path (DTAP) to be used interregionally. Other objectives are: to define the prevalence of the pathologies (both symptomatic and asymptomatic, both in adults and children) and to improve collaboration between the patient Associations and the Physicians.

On behalf of the Interregional Piemonte and Valle d'Aosta "Chiari and Syringomyelia" Consortium.

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Introduction

According to the European Union Actions Plans, the Rare Diseases (RD) are life-threatening, chronically disabling pathologies, with such a low prevalence requiring a spe- cialists joint collaboration to prevent a significant pre- mature mortality or morbidity, i.e. a relevant reduction in the quality of life or in the socio-economical potential of the affected persons [1]. In Europe the chosen threshold value was lower than one individual out of 2,000 to define a disease as Rare. With this criteria 5-8,000 different RD have been diagnosed out of 15 million of people in Europe alone. The diagnosis and care for RD has been indicated as priorities for Public Health by the Italian National Health Plan (1999); in 2001 a specific National Law (Ministerial Decree 279/2001) set up the Italian National Network for RD to deal with the prevention surveillance, the diagnosis and the treatment of the RD [2]. The same law activated the RD National Register located at the National Institute of Health (NIH), which is expected to receive epidemiological data from regional centres. The DM 279/2001 determines the RD that have right to ticket exemption, e.g. Arnold- Chiari Syndrome (code RN0010) for the symptomatic Chiari Malformation (CM).

Whilst the prevalence of Syringomyelia is known to be 8.4:100,000 [3], there are no available prevalence data for CM in Italy, because the case collection has started recently. A Regional Decree (2-03-2004, n. 22-11870) stated the Regional Network for the prevention, screening, diagnosis and therapy of Rare Diseases. Moreover it obli- ges the Regional Hospital Units to register RD in the Regional Network and sets up the Regional Centre for the Coordination of Rare Diseases [4]. A second decree (Regional Decree, 12-04-2005, n. 38-15326) sets up a Specialistic-Technical Committee to work in cooperation with the Regional Centre for the Coordination of RD and confers the benefits of the Ministerial Decree 279/2001 to about fifty RD [5], including Syringomyelia-Syringobulbia (exemption code RF0321). In fact, Syringomyelia is recognised as RD only in a few Italian regions, such as Piemonte, Valle d'Aosta, Toscana and Marche.

The Piemonte model of "diffuse" network is unique in Italy: it involves all the regional Health System specialists in the diagnosis, therapy and, above all, in the follow up and treatment. A crucial point of this model is carried out by the Consortium Activities, multidisciplinary groups, operating throughout the Region, composed by volunteer Physicians, Biologists and health care workers; they form working groups to enhance the research and the develop- ment of shared protocols for a standardized regional diagnostic-therapeutic approach.

In 2008, the Consortium Activities were approved with the aim of developing a dedicated, interdisciplinary and shared interregional diagnostic-therapeutic assistance path (DTAP); the next step will be to define the prevalence of Syringomyelia and Chiari in Piemonte and Valle d'Aosta, with special reference in the classification in symptomatic and asymptomatic classes, that may have a quite different outcome and consequent needs for heath-care.

Materials and methods

The Interregional Piemonte and Valle d'Aosta CSC was promoted in Torino by Multidisciplinary Group for Chiari and Syringomyelia at CTO Hospital: this specialist team involves many branches of medicine and care (neurolo- gists/neurophysiologists, neurosurgeons, neuroradiologists, physiatrists, spinal-surgeons, foniatrists, neuro-urologists, pain specialists, psychologists and social workers, dedi- cated nursing).

Its activities include the confirmation of the diagnosis and the patients selection for surgery, the planning of periodical Multidisciplinary Group evaluations both the surgical and non-surgical cases, the pre-surgical setup and post-surgical follow up and the identification of diagnostic- therapeutic protocols DTAP.

The DTAP begins with the neurologist in a dedicated neurological outpatients department and involves the other specialists in various extents, depending on the course of the disease. When a patient presents with suspected or defined Chiari Syndrome or Syringomyelia, then the diagnosis is confirmed by the neurological team that cer- tifies it with an exemption request for RD. Instead, in the case of a preliminary diagnosis, a temporary exemption is supplied, to perform free diagnostic examinations further needed; after that, if the diagnosis is confirmed by the multidisciplinary team, the permanent exemption is given for free care and therapy. Whilst, if the disease is not confirmed, then the temporary exemption is not renewed.

The CSC organization has many further phases, which includes: preparing diagnostic-therapeutic criteria and dif- ferentiated protocols (symptomatic/asymptomatic; child- hood/adulthood) as well as developing an interregional, shared DTAP.

Results

In a time span of a year, 102 patients were visited at the dedicated neurological outpatients department Chiari- Syringomyelia: 81 females, 21 males; 68 patients with radiological diagnosis of Chiari Malformation (65 CM-1 type and 3 CM-2 type; 37 isolated forms); 65 with radio- logical diagnosis of Syringomyelia (57 primary and 8 sec- ondary forms; 26 isolated forms). Thirty-one patients were syringomyelia-Chiari I complex (30%). Forty-one patients (60%) with diagnosis of CM (Arnold-Chiari Syndrome, exemption code code RN0010) and 38 (58%) syringomyelic patients (syntomatic Syringomyelia, exemption code code RF0321) obtained the exemption, according to Ministerial Decree 279/2001 and Regional Decree, 12-04-2005. Meetings on Syringomyelia and Chiari were organized by CSC in Torino, Cuneo and Alessandria to share diag- nostic-therapeutic criteria, to develop interregional proto- cols, to promote group research, to improve collaboration between patient Associations and physicians, to improve the relationships with the Italian Societies for Neurology, Neurosurgery and Rehabilitation.

Discussions and conclusion

Since 2005 in Piemonte and Valle d'Aosta there is a code for the patients affected by isolated Syringomyelia that avoids them from any expense for care or medication; whilst, the Chiari Syndrome patients have the complete exemption for RD in all the nation since 2001.

In Piemonte and Valle d'Aosta Interregional Network for Rare Diseases activated the CSC, "Chiari and Syrin- gomyelia" Consortium, with the objective to obtain homogeneous diagnostic criteria, to develop a dedicated, interdisciplinary team and to share the same diagnostic- therapeutic paths in the two regions. Next step will be to define the prevalence of Syringomyelia and Chiari Malformation in Piemonte and Valle d'Aosta, with special reference to their clinical correlates (symptomatic and asymptomatic forms); moreover, it will organize health operators training programs, so enhancing the cooperation between the patients and the researchers.

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