Guidelines to be followed by centres, services and units in order to be designated as Reference Centres, Services and Units of the National Health System, as agreed by the Interterritorial Board

42. HEREDITARY ATAXIA AND PARAPLEGIA

Hereditary ataxia and paraplegia are neurodegenerative syndromes affecting the nervous spinocerebellar systems. They include a group of heterogeneous syndromes which main characteristics are the slowly progressive cerebellar ataxia (occasionally episodic ataxia or spastic paraparesis) and the primary spinocerebellar degeneration.

These are rare diseases when individually considered, although as a group require a complex differential diagnosis. The nosological situation has changed in the last years with the development of genetic diagnosis. This has allowed identifying different varieties and answer questions which the clinical-pathological phenotype could not. Until now, more than 20 spastic paraplegias have been described, as well as more than 30 progressive ataxias genetically different. Under these conditions, diagnosis of a disease within a specific family has become a complex work that requires enough experience as main condition, taking into account the low prevalence of these disorders.

Differences in prognosis among the different entities turn the genetic information into a challenge that must be based on a detailed and experienced clinical and molecular analysis. Diagnosis and therapeutic approaches to hereditary ataxia and paraplegia require health staff with experience and the ability to perform an exhaustive molecular analysis. The diagnosis of patients with progressive ataxia and paraplegia without known familiar history it is also important. Reduction on family size has raised the probability for those isolated cases, especially when transmission is autosomal recessive, complicating the diagnosis unless a systematic and experienced method is used.

Existence of reference centres with special dedication to the diagnosis of these diseases allows approaching a global, clinical and genetic diagnosis of these conditions in specific families, guiding current and future treatments, and facilitating genetic counselling. National centres might act as consultants for cases with diagnostic or therapeutic complications or needing genetic counselling.

A. Rationale for the proposal

► Epidemiological data on hereditary ataxia and	Distribution of the different types of hereditary ataxias and paraplegias varies

paraplegia (incidence and prevalence).	depending on geography and other circumstances. Their prevalence was established in Cantabria in 20 cases per 100,000 population. According to this, in Spain there would be around 10,000 patients suffering from these conditions. This prevalence varies for each subtype of the disease. As a guideline, with regard to the autosomal dominant cerebellar ataxia the most common worldwide is the SCA3 and in Europe, as well, the SCA1 and SCA2. In Spain, the most frequent is also SCA3, followed by SCA2. Regarding autosomal recessive cerebellar ataxia, the most common, worldwide, is Friedreich's ataxia. Considering an approximate incidence of 0.4 cases of hereditary ataxia per 100,000 population, and maybe a similar number for acquired ataxias, there would be 320 new cases per year in a population a little bit over 40 million people. Out of these, 30% might require assessment in a reference centre, around one hundred patients each year. Candidates would be isolated cases or hereditary cases with a nosology complicating the diagnosis.

Guidelines to be followed by Centres, Services and Units in order to be designated as Reference Centres, Services and Units treating patients with hereditary ataxias and paraplegias

► Experience of the Reference Centres, Services and Units:	
- Activity: • Number of patients (minimum and optimal) with hereditary ataxias and paraplegias that should be assisted in a year to ensure an adequate care.	- 30 new patients per year. - Monitoring of 100 patients per year.
- Other data: research on the subject, postgraduate teaching, continuing training,	- Accredited postgraduate teaching: Unit participation in the internship and residency programme of the Centre.

publications, etc.	- Participation in research projects and publications in the field[a]. - Continuing training programme standardized and authorized by the centre's board of directors. - Clinical multidisciplinary sessions, at least once a month, in order to make decisions and coordinate treatments.
► Specific resources of the Reference Centres, Services and Units:	
- Human resources required for the adequate care of hereditary ataxias and paraplegias.	- 2 neurologists with part-time, but preferential, dedication to this type of diseases coordinating neurophysiological, neuroimaging and molecular genetic testing. - Nursing staff. - Administrative staff.
Basic education of the team members[b].	- Neurologists with, at least, three year experience in treating patients and families with degenerative spinocerebellar diseases. It is recommended that they would have experience in genetic-molecular diagnosis. - Nursing staff with at least, two year experience in treating this type of patients.
- Specific equipment required for the adequate care of hereditary ataxias and paraplegias.	- Care area. - Neurology hospitalization area.
► Resources from other units and services besides those belonging to the Reference Centres, Services and Units required for the adequate care of hereditary ataxias and paraplegias.	- Clinical neurophysiology services/unit with, at least, two neurophysiologists with at least two year experience in testing patients with progressive ataxias and paraplegias, and having available electromyography, electroneurography and evoked potentials techniques. - Genetics services/unit with experience in hereditary ataxias and paraplegias. Ability to preserve samples, preferably as an organized biological bank, and directly connected to the neurologists. With experience in DNA extraction and its subsequent storage and processing. DNA bank available (and by extension, RNA and cell lines banks). It must

	have a sample channel system which occasionally might be required. - Paediatrics services/unit with experience in patients with hereditary ataxias and paraplegias. - Nuclear medicine services/unit with experience in patients with hereditary ataxias and paraplegias. - Ophthalmology services/unit with experience in patients with hereditary ataxias and paraplegias. - Cardiology services/unit with experience in patients with hereditary ataxias and paraplegias. - Imaging diagnostic services/unit with, at least, two radiologists with a minimum of two year experience in neuroradiology (MRI, CT scan and other neuroimaging studies). - Rehabilitation services/unit, with at least two year experience in patients with hereditary ataxias and paraplegias and with at least one physiotherapist dedicated to these patients. - Trauma and orthopaedics services/unit with experience in patients with hereditary ataxias and paraplegias. - Psychiatry services/unit, with experience in patients with hereditary ataxias and paraplegias. - Pathological anatomy services/unit, with experience in patients with hereditary ataxias and paraplegias. - Urology services/unit with experience in patients with hereditary ataxias and paraplegias. - Internal medicine services/unit with experience in patients with hereditary ataxias and paraplegias. - Pulmonology services/unit with experience in patients with hereditary ataxias and paraplegias. - Digestive diseases services/unit with experience in patients with hereditary ataxias and paraplegias. - Biochemistry services/unit with experience in patients with hereditary ataxias and

	paraplegias. - Microbiology services/unit with experience in patients with hereditary ataxias and paraplegias. - Immunology services/unit with experience in patients with hereditary ataxias and paraplegias. - Social workers services/unit.
► Procedure and clinical results indicators of the Reference Centres, Services and Units [c]:	**The indicators will be agreed with the Units that will be designated.**
► Existence of an adequate IT system (Type of data that the IT system must include to allow identification of the activity and evaluation of the quality of the services provided)	- Filling up the complete MBDS of hospital discharge. - The unit must have a *registry of patients with hereditary ataxias and paraplegias* which at least must include: - Medical record number. - Date of birth. - Sex. - Patient's habitual region of residence. - Admission date and discharge date. - Type of admission (Emergency, planned, other). - Type of discharge (Home, hospital transfer, voluntary, death, transfer to a healthcare centre, other.) - Service in charge of patient's discharge. - Main diagnosis (ICD-9-CM). - Other diagnosis (ICD-9-CM). - Diagnostic procedures provided to the patient (ICD-9-CM): Type of procedure and date when it was provided. - Therapeutic procedures provided to the patient (ICD-9-CM): Type of procedure and date when it was provided. - Complications (ICD-9-CM).

	- Monitoring. - The unit must have the required data which should be sent to the Spanish National Health Service Reference Centres, Services and Units Appointment Commission Secretariat for yearly reference unit monitoring.

[a] *Criteria to be assessed by the Appointment Commission.*
[b] *Experience will be accredited by certification from the hospital manager.*
[c] *Clinical results standards, agreed to by the experts group, will be assessed, initially by the Appointment Commission, while in the qualification process, as more information from the Reference Centres, Services and Units is being obtained. Once qualified by the Appointment Commission, the Quality Agency will authorize its compliance, as for the rest of guidelines.*

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