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

37. INHERITED HEART DISEASES (INCLUDING HYPERTROPHIC CARDIOMYOPATHY)

Familial cardiomyopathies (hypertrophic, dilated, restrictive, arrhythmogenic right ventricular dysplasia, spongy cardiomyopathy or noncompacted...) and channelopathies include most of the causes of sudden death in young people and are also an important cause of sudden death in older patients. All these conditions have a very heterogeneous clinical appearance and a progress difficult to predict. These are inherited diseases genetically caused, and genetic diagnosis arises common logistic and ethical problems in the execution as well as in the interpretation and communication of the results.

Specialization in this topic requires, likewise, a permanent update in order to:

- Adequately describe the prognosis.
- Ensure choosing the best therapeutic option for each case.
- Guarantee its implementation by a team with experience in the field.

These characteristics imply that adequate care for these diseases requires specific tools, broad experience, a basic-clinical multidisciplinary approach, and a critical mass difficult to attain.

The main reason for designating reference units for these pathologies is the need of an intensive dedication of the specialists (cardiologists) allowing them to follow the fast rhythm of change in knowledge about these heterogeneous conditions, especially in terms of clinical implications of genetic information.

A. Rationale for the proposal

► Epidemiological data on inherited heart diseases (incidence and prevalence).	Natural sudden death entails 13% of the deaths between the ages of 24 and 75, 90% of cardiovascular origin (8,100 deaths per year). When itemized by heart condition, around 80% are due to ischemic heart disease (first cause of sudden death from 35

	years onwards), 14% to cardiomyopathies (around 1,000 deaths/year) and in 4% of the cases autopsy and complementary studies cannot subscribe to a cause of death. Inherited heart diseases are the most frequent cause of sudden death in young people (<35 years of age), being the cardiomyopathies basically 50% of the cases. Estimated prevalence of these conditions: Prevalence of inherited heart diseases may fluctuate from 1/500 for the hypertrophic cardiomyopathy to 1/5,000 for the channelopathies. - Hypertrophic cardiomyopathy: 1/500 in the general population, being a condition with a high clinical and genetic heterogeneity, with multiple variations, all of them rare (more than 600 different mutations in more than 15 genes associated to the disease). - Familial dilated cardiomyopathy (genetic cause): 1/2,000-3,000. - Restrictive cardiomyopathy: <1/5,000. Very high morbimortality. - Arrhythmogenic right ventricular dysplasia: 1/2.500-3,000. Significant morbimortality in young people (sportspeople). - Noncompacted cardiomyopathy: 1/5,000-10,000. - Brugada syndrome: 1/200-2,000. High mortality in young people. - Long QT syndrome: 1/5,000. High mortality in children and teenagers. - Short QT syndrome: 1/10,000. Very high mortality. - Catecholaminergic polymorphic ventricular tachycardia: 1/10,000 population. Very high mortality in young people (sportspeople)
► Data on the use of diagnosis and therapeutic procedures.	Special diagnosis procedures: - Genetic studies: A minimum of 1,000 patients per year may benefit from a genetic study in Spain. This number will rapidly increase according to improvement in the knowledge of genetic basis and of the correlation genotype-phenotype of these conditions, as well as the use of this studies for diagnosis of sudden death and idiopathic syncope spreads out. A percentage of these patients (between 5-10%) will require implantation of an automatic defibrillator. - Imaging studies: Echocardiography in these patients requires experience in the

	interpretation of emergent signs and in differential diagnosis. MR is a very useful technology for these conditions when in expert hands. - Electrophysiological studies: Indispensable for diagnosis of channelopathies. Therapeutic procedures: - For hypertrophic cardiomyopathy: Myectomy: 25-50/year in Spain. Alcohol septal ablation: 80-120/year in the country. Implantable defibrillator: 100-200/year. - For dilated cardiomyopathy: Heart transplantation: Idiopathic dilated cardiomyopathy is the cause of 50% of the transplantations performed. For channelopathies and arrhythmogenic right ventricular dysplasia: Implantation of automatic defibrillator: 100-200/year.

B. Guidelines to be followed by Centres, Services and Units in order to be designated as Reference Centres, Services and Units for the care of inherited heart diseases

► Experience of the Reference Centres, Services and Units: - Activity: • Number of patients that should be assisted in a year to ensure an adequate care of inherited heart diseases.	- 75 new patients diagnosed with inherited heart diseases per year. - 150 relatives assessed per year.
• Number (minimum and optimal) of techniques, technologies and procedures similar to those specific to the designation requested that should be performed in a year.	- Diagnosis or therapeutic techniques available in the hospital that are performed to patients with inherited heart disease include study of new cases, follow-up of patients and family studies (during the first visit and follow-up): ◆ 250 echocardiographies, 50 cardiac MR, 100 Holters, 100 stress tests. ◆ Ability to perform pharmacological stress test (flecainide, procainamide or

	adrenaline). ◆ Ability to perform genetic studies in all the index patients.
- Other data: research on the subject, postgraduate teaching, continuing training, publications, etc.	- Accredited postgraduate teaching: Unit participation in the internship and residency programme of the Centre. - 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. - Promoting training courses on these pathologies for health professionals.
► Specific resources of the Reference Centres, Services and Units:	
- Human resources required for the adequate care of inherited heart disease.	- 2 cardiologist with full-time dedication to inherited heart diseases. - Nursing staff.
- Basic education of the team members[b].	- Cardiologists with 3 years experience in inherited heart diseases. - Nursing staff with more than 2 years experience in cardiologic tests and family tree drawing.
- Specific equipment required for the adequate care of inherited heart disease.	Unit for inherited heart disease: - Inherited heart disease programme authorized by the centre's director. - Clinical practice with specific dedication to inherited heart diseases: Fundamental to obtain the required knowledge and experience for treating these problems and adequately coordinate the required care procedures.
► Resources from other units and services besides those belonging to the Reference	- Diagnostic Imaging services/unit, with experience in performing procedures in inherited heart conditions, including high end echocardiography and MR, with ability

Centres, Services and Units required for the adequate care of inherited heart disease.	to perform studies of inherited heart diseases. - Electrophysiology and arrhythmias unit with experience in inherited heart diseases, including provocation-exposure techniques, such as the Flecainide text in the Brugada syndrome. - Genetics services/unit with experience in inherited heart diseases. - Hemodynamics unit (diagnosis and therapeutic procedures) with experience in inherited heart disease, including alcohol septal ablation. - Heart surgery services/unit with experience in inherited heart diseases. Including performing myectomies for the treatment of the hypertrophic obstructive cardiomyopathy. - Pathological anatomy services/unit, with experience in inherited heart disease. - Paediatrics services/unit with experience in inherited heart disease. - Active heart transplantation programme and authorized according to the Royal Decree 2070/1999, December 30th, establishing the general basis for clinical harvesting and use of human organs and the territorial coordination in donation and transplantation of organs and tissues, allowing for response and continuity to the needs arising from the treatment of patients with inherited heart disease.
► 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 inherited heart diseases* which at least must include: - Medical record number. - Date of birth. - Sex. - Patient's habitual region of residence. - Date when assisted or admission date and discharge date.

	- Type of care or 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. - Family history, known heart disease, phenotype, known genotype in the family, etc. - 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). - Patient monitoring: adverse events. - 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 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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