

ICORD 2014 Annual Meeting

“An integrative model of rare diseases management in a tertiary hospital: from basic research to clinical practice and beyond”



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Barcelona, Spain

Ede, The Netherlands- 8th of October 2014



AFFECTED POPULATION

In Rare Diseases

USA: 25 milion (8.4%)

UE-27: 29-36 milion (6-8%)

SPAIN: 3 milion

CATALONIA: 300.000 - 400.000

Rare Disease in Europe: **1 /2,000 people (<5 individuals / 10,000 inhabitants) (prevalence)** (*EC Regulation on Orphan Medicinal Products*).

It is estimated there are between **6,000 y 8,000** rare diseases.

In Europe, when the disease is either **life-threatening or chronically incapacitating and it requires a merged effort to treat it**, it is also classified as rare disease.

They usually affect **familiar and social environment** and they require multidisciplinary therapeutic approach with a high economic cost.

80% of rare diseases have a **genetic origin** and they affect both physical and psychological development. There are also rare **infections**, rare **cancers** or rare **degenerative** diseases.



WHAT SHOULD BE DONE TO IMPROVE THIS SITUATION ?

- **Improve the understanding of rare diseases** by society and people working with them by offering high quality information
- **Develop appropriate public politics**
- Increase **international collaboration** in this area
- **Increase our knowledge of all rare diseases** and not only the 'most frequent' ones
- Develop **new diagnosis methods and new treatments**
- **Promote the association of groups of patients** in order to share their experiences and get better health practices.



Nowadays, RD is a **priority international collaboration field**.

Singular initiatives exist, especially in the research area:

- Europe: **Eranet E-Rare** and **7 PM**
- World: **International Rare Diseases Research Consortium (IRDiRC)**

Estrategia en Enfermedades Raras del Sistema Nacional de Salud

Estrategia aprobada
por el Consejo Interterritorial
del Sistema Nacional de Salud
el 3 de junio de 2009

SANIDAD 2009
MINISTERIO DE SANIDAD Y POLÍTICA SOCIAL

**Approval
3rd June 2009**

NATIONAL STRATEGY IN RD (NATIONAL HEALTH SYSTEM)

Health related assistance in Spain is transferred to the Autonomous Community. The Health Ministry acts as coordinator, manager and regulator of the overall system, although it does not provide direct health services.

The **National Strategy in Rare Diseases from the National Health System** was approved on the 3rd of June 2009 by the **interterritorial board of the National Health System**. It is a document elaborated by the administration, the autonomous communities (regions), scientific societies and other experts.

PHASES OF STRUCTURATION :

- **1st PHASE: General aspects** including justification, strategy goals and situation of RD in Spain;
- **2nd PHASE: Development of 7 strategy lines with objectives and recommendations for each one of them;**
- **3rd PHASE: Evaluation and follow-up process.**



CAAM

In 2009, the *Servei Català de la Salut* (CatSalut; Catalan Health Service), created:

Rare Diseases Committee (Comisión Asesora en Enfermedades Minoritarias; CAMM)

OBJECTIVES:

- **Improve services and performances** on RD, with good **early diagnosis** and the establishment of a **network of reference centers services**, units and skilled professionals;
- **Propose** actions to enhance **coordination between different levels of care and foster cooperation between the various stakeholders** including Catalonia's government, organizations affected by rare diseases, health professionals, pharmaceutical and medical devices and the research sector;
- Creation of CATFAC (Consell Assessor of Tractaments Farmacològics d'Alta complexitat), that **regulates the administration of treatments** to be dispensed by the specialized centers with recognized expertise professionals.



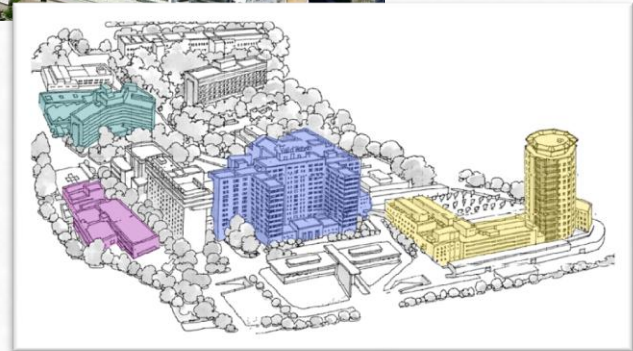
In July 2010 CAMM approved the **Healthcare Model in Rare Diseases in Catalonia** that it is currently valid.

Two differentiated levels are established:

- **Specialized healthcare with strong expertise in RD** that has been formed by Complex Centers and tertiary reference hospitals in Catalonia.
- **Proximity or community healthcare**, constituted by the primary care services, by the specialized referential care (secondary care) and by the overall public resources from the education and social proximity areas from the patient's home.



- **1952** | Inaugurated in Barcelona;
- **2009** | Accredited by *Instituto de Salud Carlos III* as *Instituto de Investigaciones Sanitarias*
- **2013** | Re-accredited by ISCiii
- Nowadays it is the Public Centre with more activity in healthcare and research in Catalonia



2012 Budget: 590 M€



7,000
professionals



1,146
beds



381
consultations



45
operating rooms

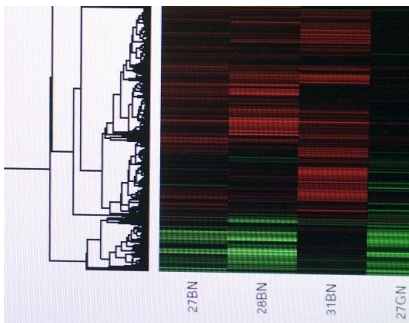
There are 5 big areas:

- General (adults)
- Pediatrics, gynecology and obstetrics
- Traumatology & Rehabilitation
- Training
- Research



ADVISORY COMMITTEE IN RARE DISEASES AT HUVH

- **Develop a Comprehensive Care Program** for patients with a RD in the Hospital, according to the model defined by the Department of Health;
- **Establish a network of RD experts** in the hospital that can integrate into other existing networks in Catalonia, Spain or at an international level, and to **ensure the diagnosis, assessment, monitoring and multidisciplinary treatment** of those affected and their families ;
- **Simplify the healthcare circuits** (also called "healthcare pathways") of the patients suffering from a RD, while ensuring good coordination and continuity of care from the hospital (especially in the transition between childhood and adulthood) and primary care in the territory served by the hospital;
- **Develop and implement healthcare protocols** to favour screening, early genetic and clinical diagnosis and treatment and efficient monitoring of patients.

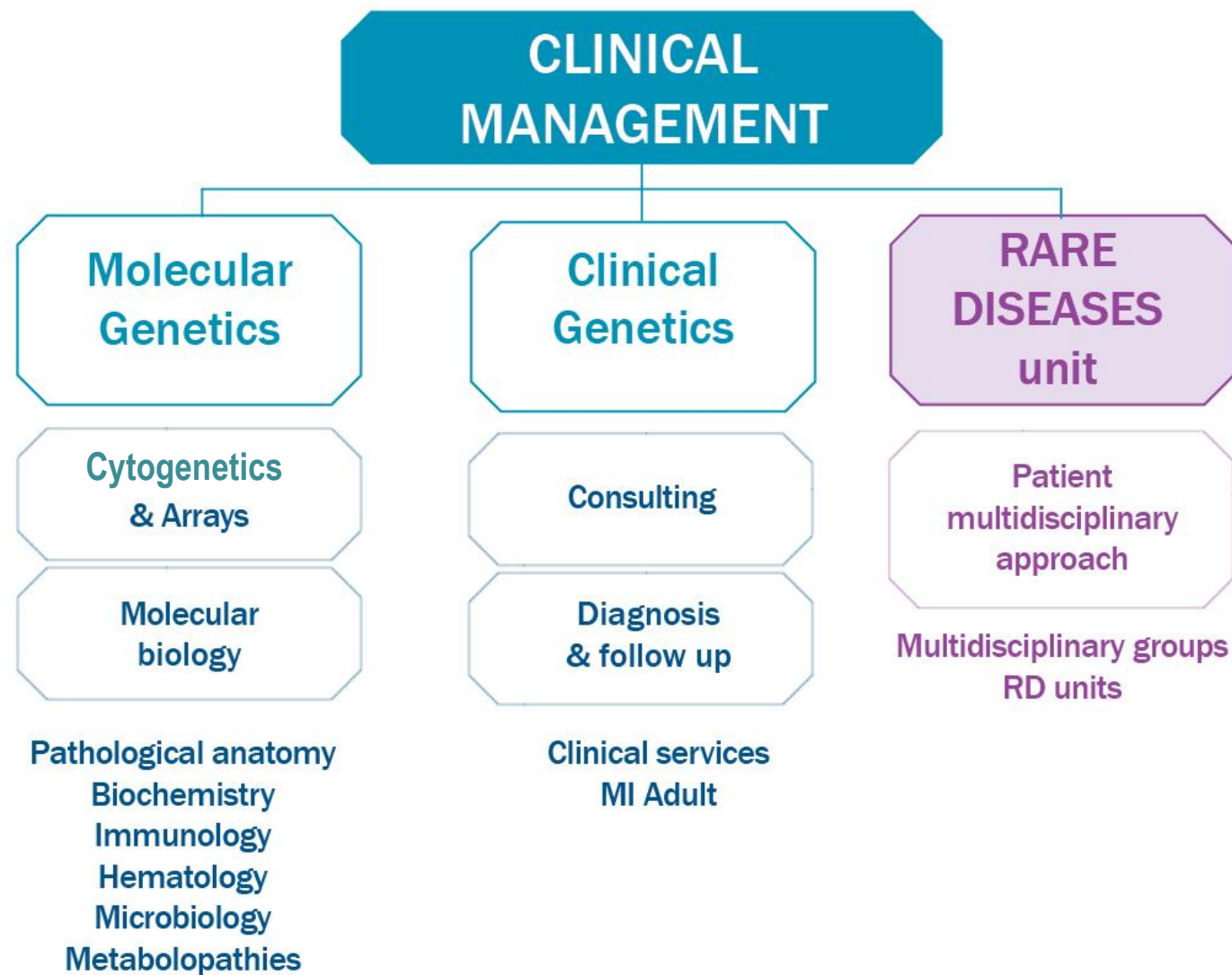


- **Develop** a set of **indicators** to **monitor and evaluate the healthcare given** to the patients and the strategies used to implement them;
- **Promote training and information activities**, and raise awareness among health professionals;
- Enhancing **basic, clinical, translational and epidemiological research in RD**;
- Processing information to the Department of Health for **epidemiological monitoring**, have a better understanding of the history, identifying the needs of these groups and assess the impact of displayed performances;
- Advise, recommend and report on scientific, technical and economic relevance of incorporating **new diagnostic tests and new therapies**;
- **Ensure the flow of communication** between the committee, the Hospital management and professionals involved in both the detection of problems and suggestions for improvement.



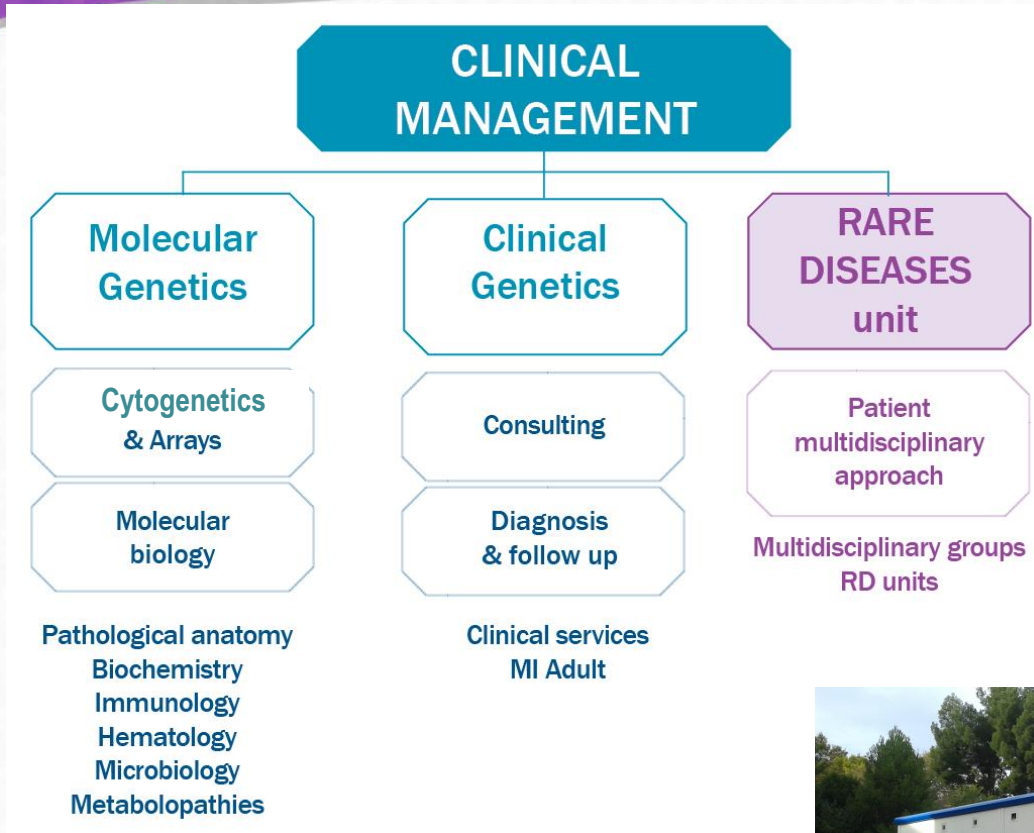
1. Pediatric Cardiology, 9
2. Pediatric Neurology, 6
3. Plastic Surgery and burned, 4
4. Bioengineering orthopedic and pediatric surgery, 2
5. Genetics, 5
6. Digestive physiology and pathophysiology, 3
7. Intensive medicine, infectious diseases & pneumology, 3
8. Endocrinology, 6
9. Autoimmune and systemic diseases 12
10. Pediatric Immunology, 4
11. Microbiology & Infectious, 1
12. Neurovascular, 3
13. Respiratory Immunology, 4
14. Neurology, 11
15. Hematology, 1
16. Pneumology, 17
17. Neurotrauma, 5
18. Mitochondrial and neuromuscular pathology, 7

TOTAL: 103 HEALTH PROFESSIONALS



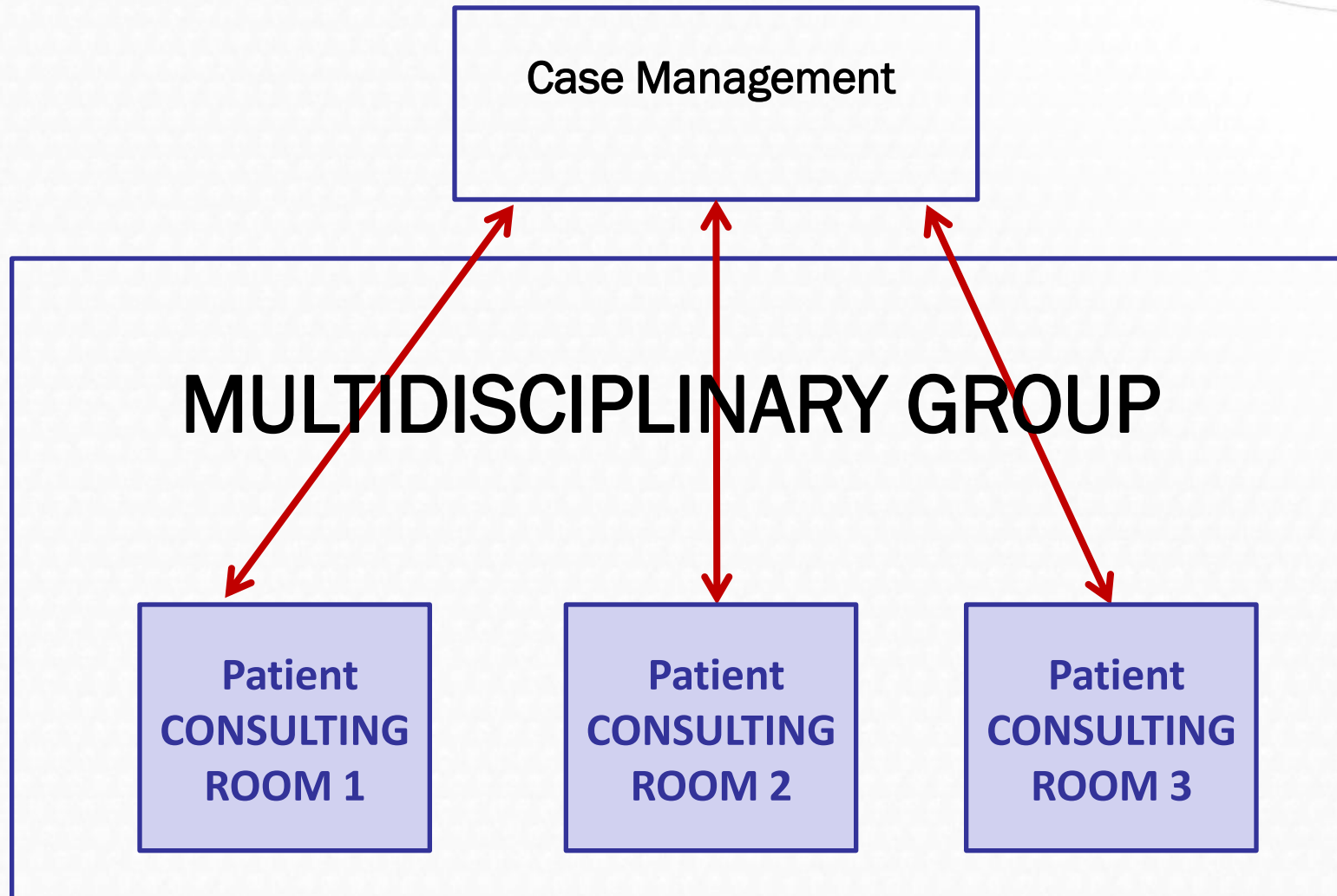
- Integrating the practice of clinical genetics (diagnosis, counseling, possible therapies)
- Integration and coordination of molecular genetic studies (healthcare and research)
- Integration and coordination of multidisciplinary monitoring of patients diagnosed with a RD (Management case-patients-doctors).
- Scale economy and sustainability
 - Multidisciplinary approach-extension casuistry
 - Adequacy of the diagnosis tests, internalization and external tests control
 - Adaptation of treatments and protocols
- Organization of teaching / training programs (Genetics specialization is ongoing)
- Develop translational research

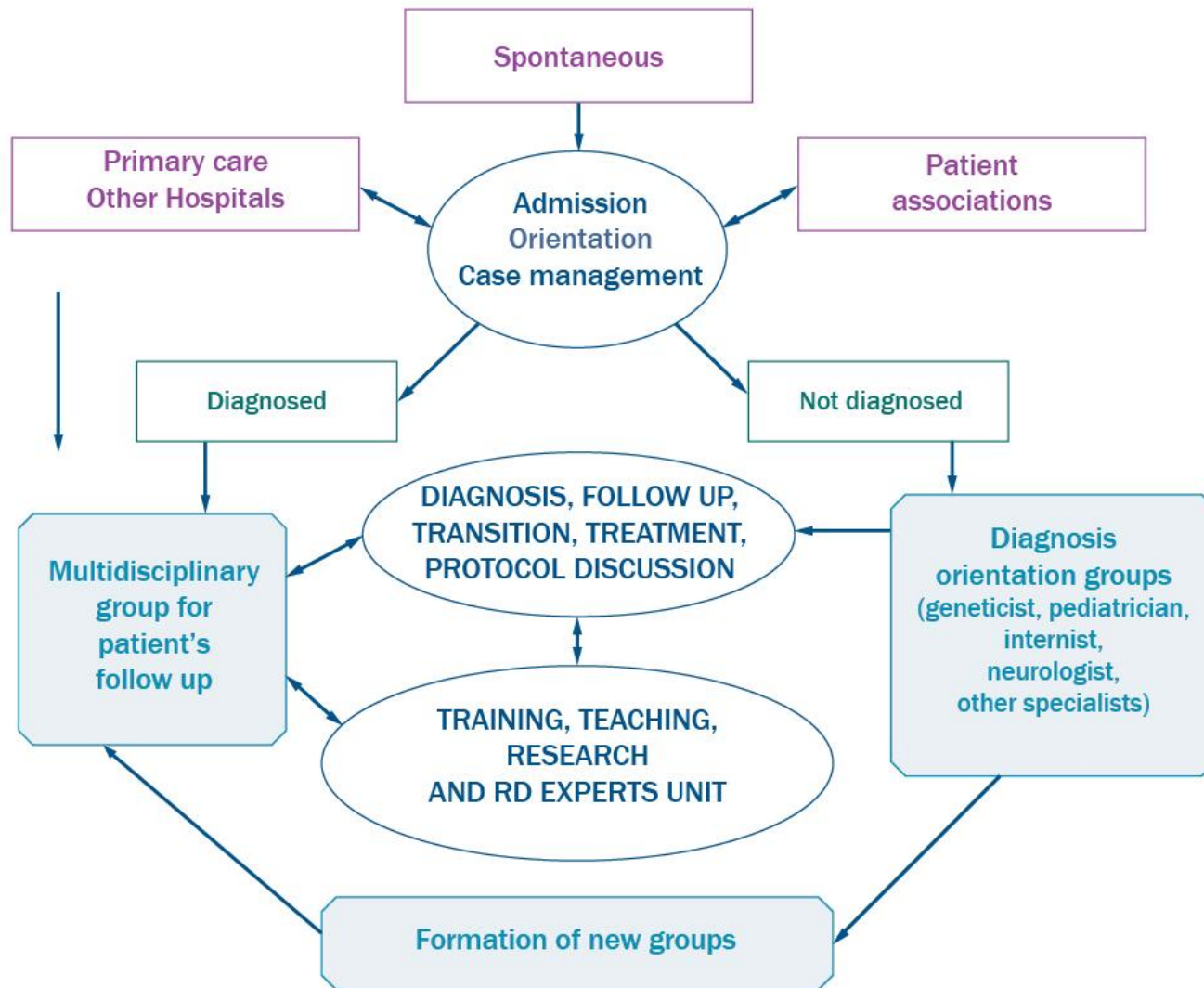
Organization of the Area of Clinical and Molecular Genetics and Rare Disease Unit



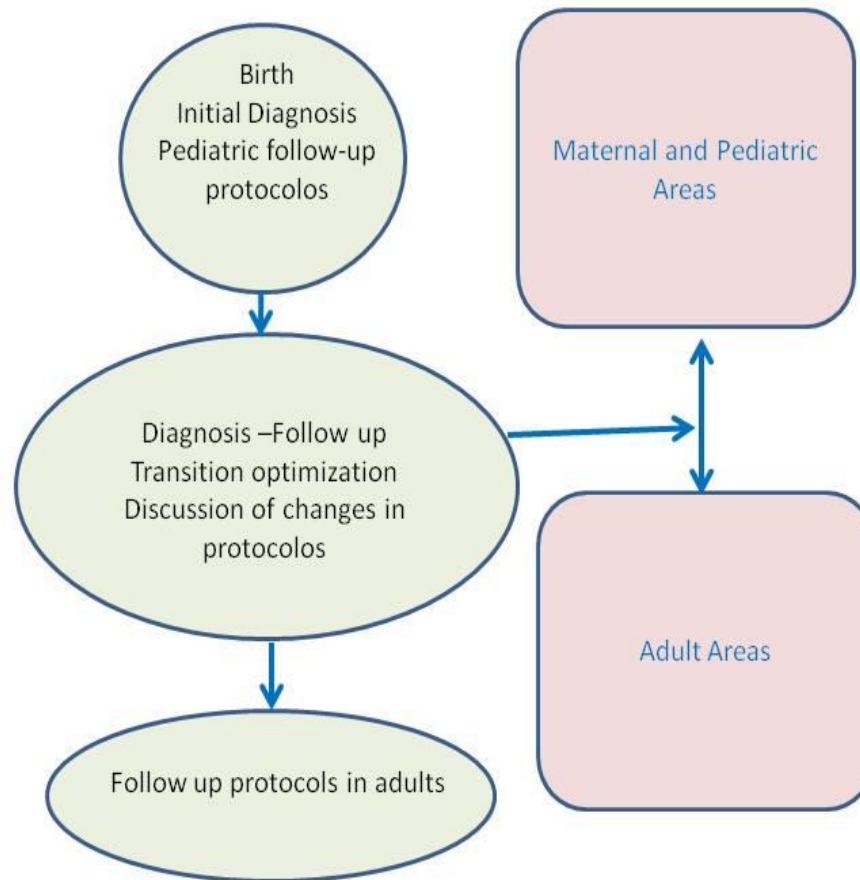
Remodeling of the Clinical Genetics & Rare Disease Units: Offices, waiting room, meeting room, rotating-residents and multidisciplinary healthcare area in Rare Diseases distributed on two floors (500m2 in total).







VH is able to offer a continuous follow up and treatment of patients diagnosed with RD from their birth up to adulthood.



191 Rare Diseases

26 Clinical areas

103 Medical doctors

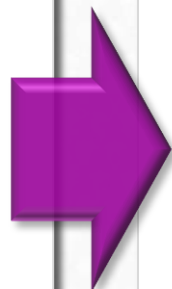
26 MIR (Residents)

46 PostDocs

62 Nurses

53 PreDocs

78 Technicians



10.5 M€ Global funds

8.5 M€ from **123** Projects

2.0 M€ from **243** Clinical Trials

571 Publications

2597.79 Accumulated IF

4.56 Average IF



THANK YOU FOR YOUR ATTENTION

AN INTEGRATIVE MODEL OF RARE DISEASES MANAGEMENT IN THE VALL D'HEBRON TERTIARY HOSPITAL AND INSTITUTE OF RESEARCH

FROM BASIC RESEARCH TO CLINICAL PRACTICE AND BEYOND

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³ CIBERER (Centre for Biomedical Network Research on Rare Diseases, ISCIII)

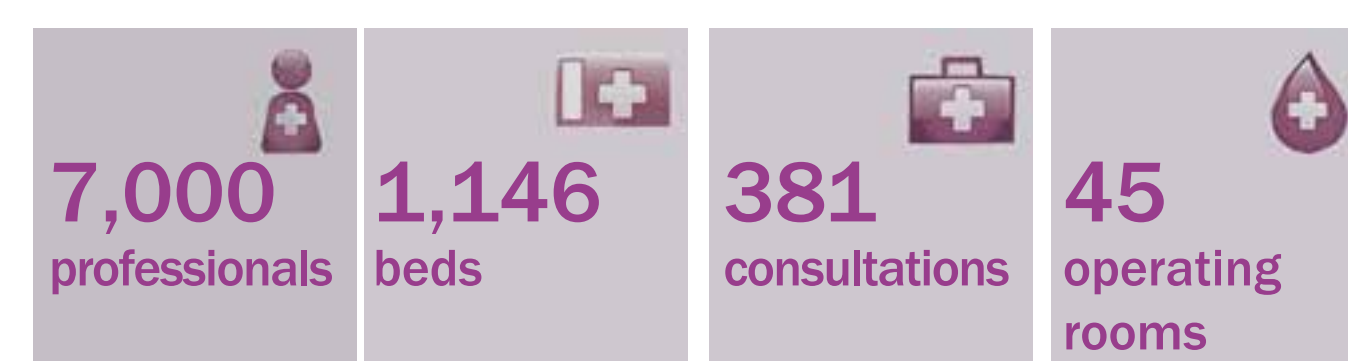
Vall d'Hebron University Hospital and Institute of Research

THE HOSPITAL

2012 budget: 590 M€

There are 5 big areas:

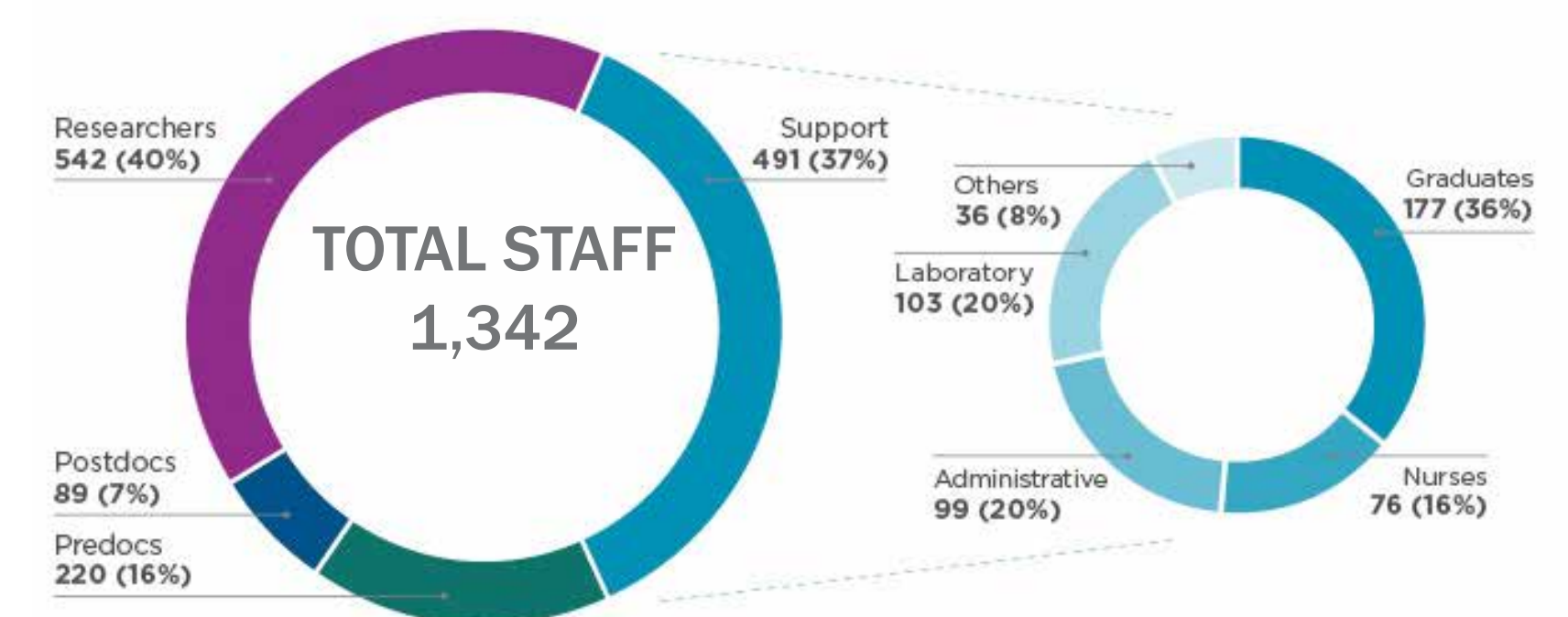
- General (adults)
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- Research



RESEARCH

At present, the 'Vall d'Hebron University Hospital and Institute of Research' is the Public Centre with the highest activity in healthcare and research within Catalonia

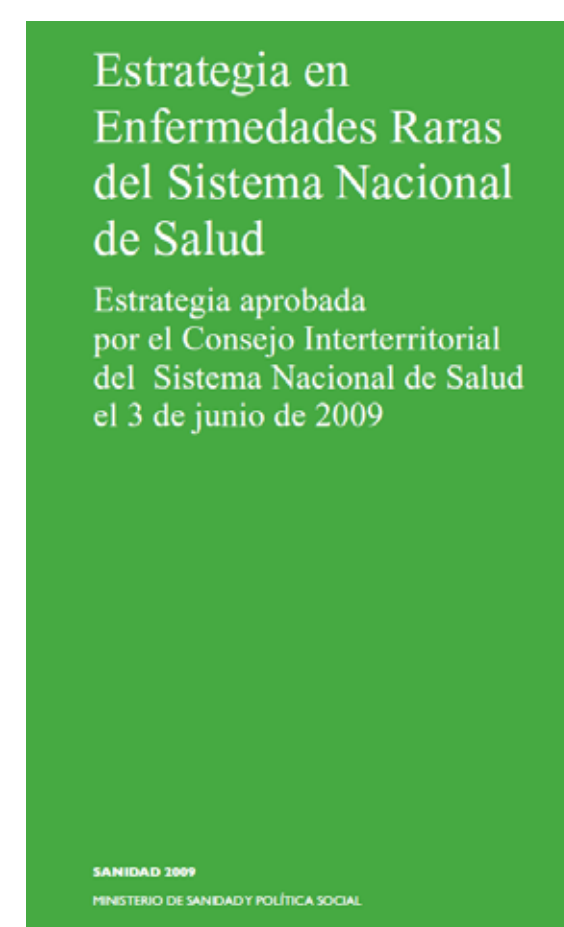
75% of our PIs are clinicians



AFFECTED PEOPLE WITH A RARE DISEASE

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Rare Diseases in Spain



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STRUCTURE

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General aspects: justification, strategic goals and situation of RD in Spain.

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3rd PHASE:
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Rare Diseases in Catalonia

Rare Diseases Committee (Comisión Asesora en Enfermedades Minoritarias, CAMM)

Created in **2009** by CatSalut, the Catalan Health Service.

OBJECTIVES:

- The improvement of RD services, including a better early-diagnosis and establishment of a network of excellence centres.
- The promotion of the coordination between different levels of assistance and the fostering of cooperation within various departments.
- The creation of CATFAC (Advisory council for Highly Complex Pharmacologic Treatments).



Most relevant Rare Diseases treated at the Vall d'Hebron Hospital and Institute of Research

► SYSTEMIC DISEASES

Systemic lupus erythematosus, **650** patients
Scleroderma, **450** patients
Sjögren's syndrome: **500** patients

► PEDIATRIC NEUROLOGY

(metabolism congenital disorders)
Lysosomal storage disorder, **28** patients
Mitochondrial disease, **20** patients (neuromuscular disorders)
More than **171** evaluated cases, since 2000



► PEDIATRIC PNEUMOLOGY

Cystic fibrosis **500** diagnosed patients

► Pediatric Tumors **2631**

► Marfan syndrome **219**

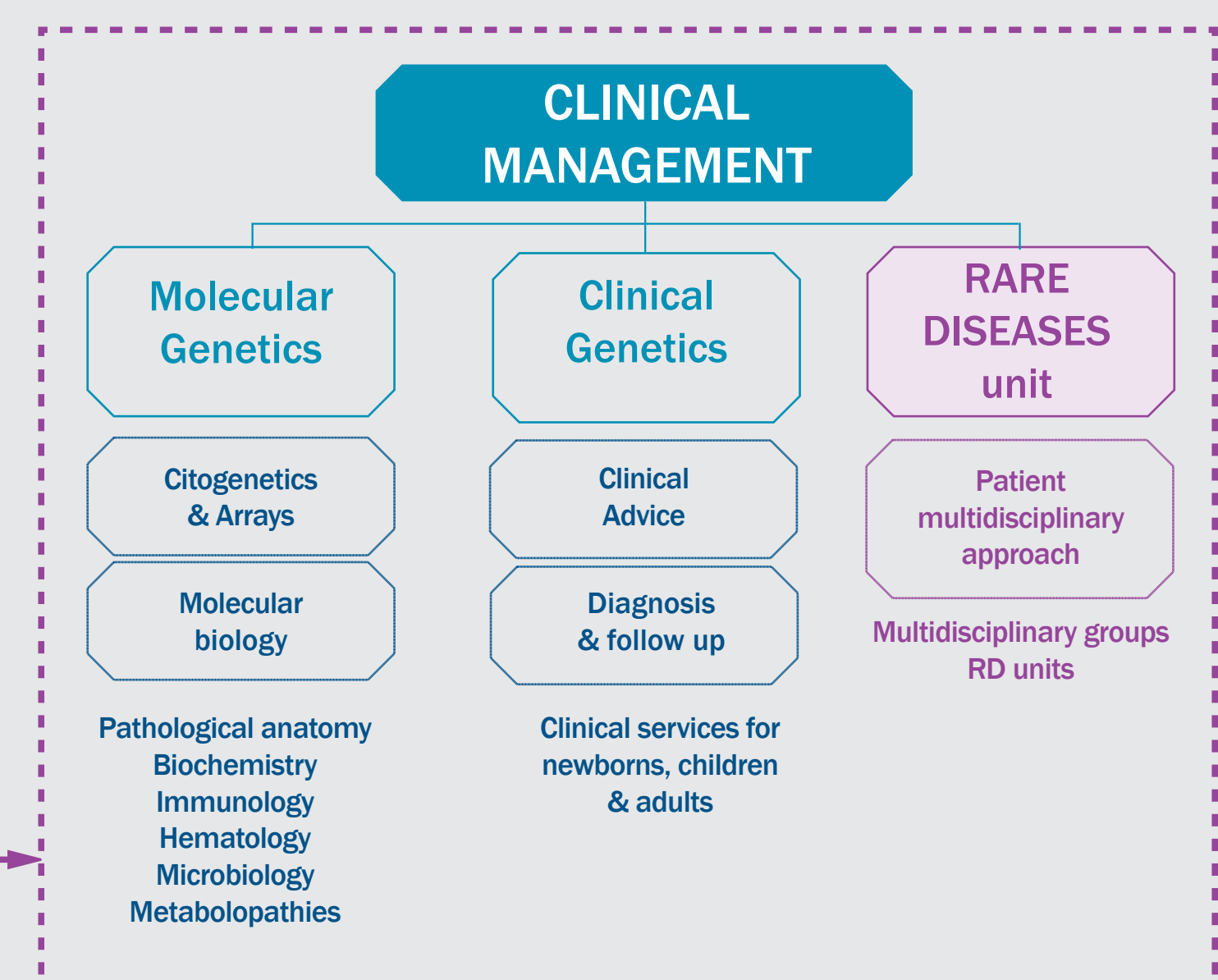
► Spina bifida **500** patients

RARE DISEASES FACTS AND FIGURES

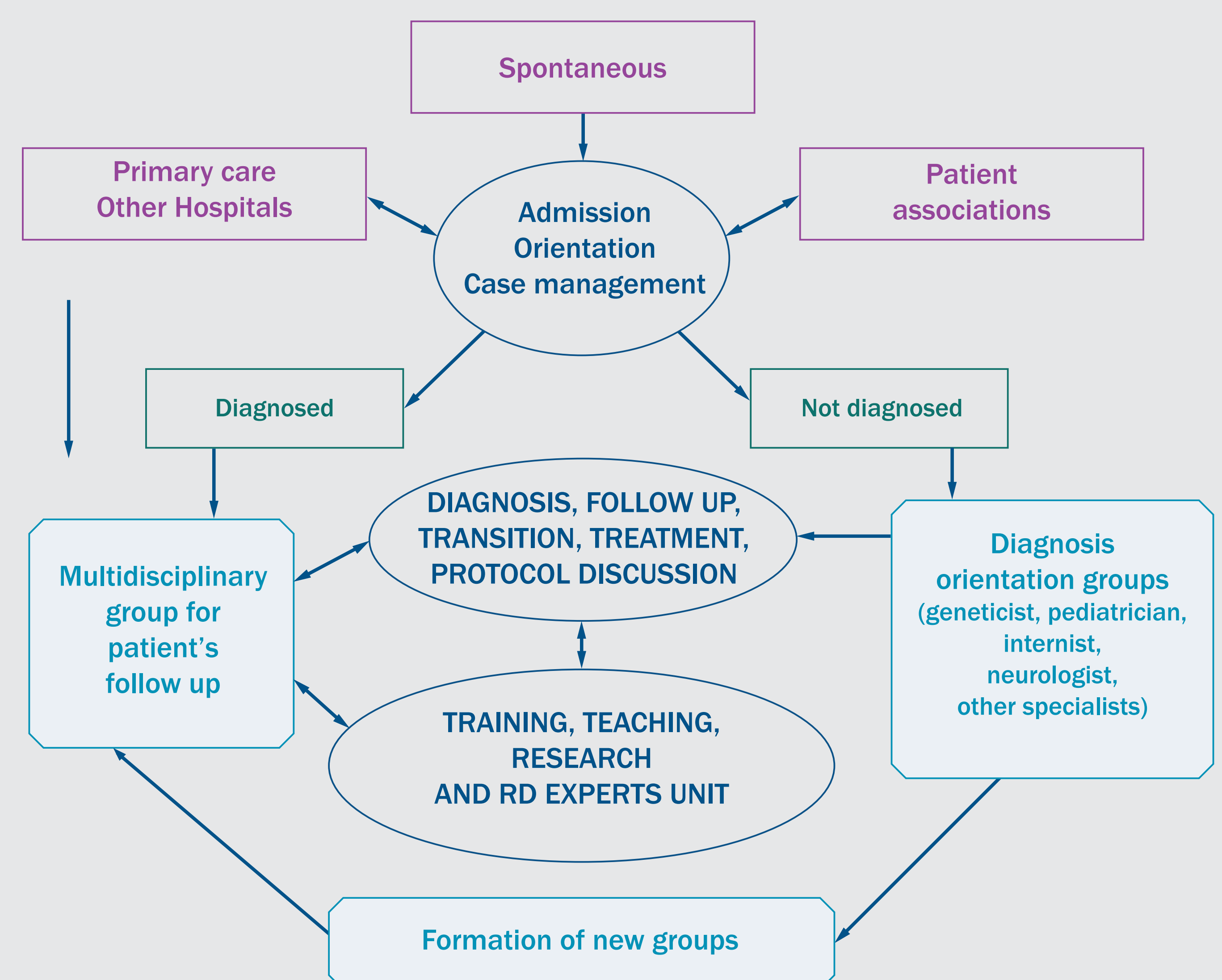
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Organization of the Clinical Genetics, Molecular Genetics and Rare Diseases Units



Patients follow up at the Rare Diseases Unit



Transition of patients to Rare Diseases Unit

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