

## EXPERTISE CENTRE FOR HEREDITARY METABOLISM DISEASES



**Department: Prof. Brigitte CHABROL** 



## Looking for FULL-TIME HOSPITAL PRACTITIONER

The paediatric neurometabolism service (CRMR hereditary metabolic diseases) at the children's hospital, CHU Timone, is currently recruiting a full-time hospital practitioner.

Applications should be sent to Professor Brigitte Chabrol: brigitte.chabrol@ap-hm.fr

The Paediatric Neurometabolism Department is accredited by **5 expertise centres** (CRMR) and **2 competency centres** (CCMR) for rare diseases:

- the CRMR for **hereditary metabolic diseases, CoMMet** (national coordinating site under the responsibility of Prof Chabrol)
- the CRMR for **mitochondrial diseases from children to adults, CALISSON** (constitutive site under the responsibility of Prof. Chabrol)
- the paediatric part of the CRMR for **neuromuscular diseases** (under the responsibility of Prof. Chabrol)
- The CRMR for **intellectual disability and rare polyhandicap** (national coordinating site under the responsibility of Pr Milh)
- The paediatric section of the CRMR for rare epilepsies (under the responsibility of Prof. Milh)
- The CCMR for children with neurofibromatosis
- The CCMR for rare inflammatory diseases of the brain and spinal cord

The paediatric neurometabolism department's medical team includes 2 university professors and 5 full-time hospital practitioners.

The paediatric neurometabolism department comprises:

- A 14-bed inpatient unit
- A 6-place neurometabolic day hospital unit
- A multidisciplinary consultation unit

## The role within the CRMR Hereditary Metabolic Diseases:

- Diagnostic, follow-up and management consultations
- Ensuring patient care
- Participation in the activities of the PACA/Corsica Regional Neonatal Screening Centre (CRDN)
- Participation in local and national multidisciplinary consultation meetings (RCP), RCP on pre-indications for the AURAGEN platform on which we depend
- Participation in the activities of the CRMR:
  - > Expertise: participation in the drafting of national protocols for diagnosis and care (PNDS); participation in the implementation of Patient Therapeutic Education (ETP) programmes; providing information for the national database on rare diseases (BNDMR)
  - Research: participation in clinical studies, updating of disease-specific registers and publications
  - > Training and information: participation in the training of medical interns and assistants in particular

**Environment:** close-knit team, friendly nursing and paramedical staff

Strengths: A paediatric neurometabolism department with regional, national and european recognition