

Competence Centres

Since 2007, the Reference Centre for Rare Skeletal Disorders has run a network of 14 recognised Competence Centres in France.

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Patient Organisations

The Reference Centre for Rare Skeletal Disorders works closely with the patient organisations concerned:

AMEM: Hereditary Multiple Exostoses

APPT: Little People

AOI: Osteogenesis Imperfecta

A la Volette Multiple Epiphyseal Dysplasia

Association Ollier-Maffucci Europe: Ollier-Maffucci

Association Syndrome de Poland: Poland syndrome

Hypophosphatasia Europe: Hypophosphatasia

FOP France: Fibrodysplasia Ossificans Progressiva

SOLHAND: Rare disease and disability support association

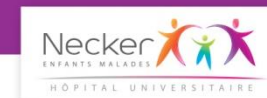
VML: Lysosomal Storage Disorders



INFORMATION

French Reference Centre for Rare Skeletal Disorders

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What are rare skeletal disorders?

Rare skeletal disorders are various conditions caused by defects in genes encoding certain proteins involved in bone growth and development. They lead to short stature and varying degrees of bone deformities.

There are more than 400 skeletal disorders, some of which are more common, such as achondroplasia, osteogenesis imperfecta and hereditary multiple exostoses. The Reference Centre for Rare Skeletal Disorders follows-up over 4,500 patients, from childhood to adulthood, and is a network of expertise in rare bone diseases.

Hospital sites in Paris

The Reference Centre is located at 4 sites within Parisian hospitals and includes:

1 Coordinating site

- **Hôpital universitaire Necker-Enfants malades**
Pr. Valérie Cormier-Daire and Dr. Geneviève Baujat

3 Constitutive sites

- **Hôpital Armand-Trousseau**
Dr. Véronique Forin (paediatric)
- **Hôpital Cochin**
Pr. Christian Roux (adult)
- **Hôpital Lariboisière**
Pr. Martine Cohen-Solal (adult)

The Reference Centre's Missions

- To establish a diagnosis
- To organise follow-up and prevent complications wherever possible
- To ensure continuity of care from childhood to adulthood
- To disseminate knowledge and good medical practice
- To take part in developing clinical and basic research programmes to study these disorders
- To participate in epidemiological surveillance
- To run and coordinate networks of medico-social and educational contacts
- To act as a point of contact for oversight bodies and patient associations
- To offer appropriate psychological and socio-educational support

Diagnosis and care

- **Diagnosis** is based primarily on physical examination and radiological investigation.
- **Patient care is comprehensive, including:** the child's growth and development, sleep, integration at school, treatment of orthopaedic complications, pain, ear nose & throat, respiratory, ophthalmological or other complications.
- **Multidisciplinary care** is essential. There is often no specific treatment for individual disorders, but the follow-up plans for each disorder are now more standardised and new treatments are currently being developed.

The main achievements since 2004

- Providing specialist antenatal and postnatal diagnosis consultations
- Producing treatment guides
- Organising specialist multidisciplinary consultations:
 - Orthopaedics & Genetics (Necker)
 - Rheumatology & Genetics (Necker)
 - Adolescent / adult transition (Cochin and Lariboisière)
- Setting up routine molecular diagnosis for more than 150 skeletal dysplasias
- Certification of specialist centres and running a network of 14 regional competence centres
- Partnerships with the biopharmaceutical industry on clinical and treatment protocols
- Organising skeletal dysplasia meetings:
 - weekly clinical/molecular medicine meeting
 - quarterly joints/skeletal dysplasia and respiratory medicine/skeletal dysplasia meetings
 - monthly radiology/skeletal dysplasia meeting
 - twice-yearly meeting between the expert centre and the specialist centres
- Creation of a university diploma in skeletal dysplasias in 2010
- Publications: 400 scientific articles; 28 chapters in reference works.

