# Referral Centre for Developmental anomalies and malformation syndromes (clinical genetics)



Rennes university hospital manages a referral centre to improve the diagnosis and treatment of rare diseases in children and adults affected by malformations and/or intellectual deficiencies.

Professor Odent, a specialist in medical genetics in the Mother and Child Centre talks to us about the objectives:

**What pathologies are concerned ?**

The official name is an acronym : ACDA, or “Accredited Centre for Developmental Anomalies”, in French: “Centre Labellisé pour les anomalies du développement”, CLAD-Ouest. These anomalies include malformations with or without the presence of intellectual deficiency. They are considered as rare diseases but they affect in total between 12,000 and 24,000 births every year. It is therefore a major public health issue which explains the large-scale involvement of a multidisciplinary team which includes doctors, care and medico-social professionals, researchers and health stakeholders in the running of the centre.

**What is a rare disease?**

A disease is considered as rare if it affects less than one person in 2000, which in France means less than 30,000 people for each disease. Nearly 7,000 rare diseases have been identified, causin variable disabilities. 80% are genetic diseases. More than 3 million French persons and 27 million European persons are affected to a greater or lesser extent by these diseases. Around fifty rare diseases affect several thousand people in France, while 500 others only affect several hundred, and thousands of other diseases affect only a few dozen people.

**Do we currently have effective therapies to treat patients affected by these malformations and deficiencies which in some case are numerous?**

For the management of symptoms yes, but unfortunately not for treating the cause of the disease, except in rare circumstances. However, enzyme therapies and therapeutic trials are beginning to appear, particularly in the field of illnesses which cause intellectual deficiency. Moreover, given the rareness and the different mechanisms of these anomalies, we do not expect the development of cure treatments such as genetic therapy to become routine. But if we cannot heal them as yet, thanks to advances in clinical knowledge and medical research, we can detect and understand them better. The identification of an anomaly enables us to provide explanations to families, to clarify genetic counselling which means the risk of recurrence, explain the principles of prenatal or preimplantation diagnosis and allows a closer relationship with patient associations. If diagnosis is made early in life, affected children can benefit not only from medical care, but also socio-educational care, which is the best for them to prevent worsening of their disability.

**The Rennes university hospital already organises clinical genetics consultations which has a high attendance rate. The referral centre is one of the driving forces.**

But all of the patients affected by genetic diseases or those who have a genetic predisposition to cancers (oncogenetics) are seen in consultations. Within the framework of the national plan for rare diseases, the ministry has created regionally and nationally accredited centres. Therefore, in 2005, it was decided to create a referral centre spread across the university hospitals of Rennes, Brest, Angers, Nantes, Tours and Poitiers and the Vannes general hospital. Practitioners from these centres have been working together for several years in particular within the Western France genetics network. A "Western Region" centre seemed like a natural progression in order to deal with the challenges of research, training of health professionals and informing families. This collaborative work is invaluable for both practitioners and patients. Indeed, the rareness of these anomalies makes them very difficult to diagnose even for highly experienced practitioners. Since 2014, the CLAD-Ouest <s>Western-ACDA</s> has been a part of the "AnDDi-Rares" network, which is the national health network for rare diseases with developmental somatic and cognitive anomalies. It gathers together 8 Centres referral for rare diseases and 7 Specialist centres for developmental anomalies, 38 molecular genetics laboratories, 44 cytogenetic laboratories, 43 foetal pathology laboratories, 40 research laboratories, 6 learned societies and 44 partner associations. It develops actions concerning patient diagnosis, monitoring and care, research, professional training and information for patients and their families.

**How does the centre actually get accreditation?**

As a coordinator, we have received financial help to recruit a hospital practitioner, a medical secretary, a coordinating nurse, a psychologist and a social worker to facilitate a multidisciplinary approach and the coordination of projects at the Western France regional scale. The advent of high-speed sequencing techniques for all genes is transforming the diagnostic approach for patients who were until now undiagnosed.