NAGEN PEDIATRICS TEAM

The NAGEN PEDIATRICS Project forms part of Navarrabiomed's **NAGEN Program**, which aims to increase knowledge and use of genome analysis technology in the Navarre public healthcare network. It is supported by the Government of Navarre's Ministry of Economic and Business Development within the framework of the Smart Specialization Strategy (S3).

The project is coordinated at the Genomic Medicine Research Unit and the initiative is led by Josune Hualde Olascoaga, a Navarrabiomed researcher and pediatrician at the Hospital Complex of Navarre.

CONSORTIUM

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MORE INFORMATION

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NAGENpediatrics

Advanced genome sequencing in pediatric patients

The **NAGEN PEDIATRICS** Project is an initiative led by the Navarrabiomed Biomedical Research Center. The project aims to perform **whole genome** analysis of pediatric patients for fast action in clinical treatment. It also includes a research area on autism spectrum disorder (ASD) in collaboration with the Group of Genomic Medicine at the Galician Foundation of Genomic Medicine.

Genetic disorders have a significant impact in pediatrics: 2% to 3% of newborns have some form of congenital anomaly and half of them are cases of genetic origin.

NAVARRABIOMED CENTRO DE INVESTIGACIÓN BIOMÉDICA

Gobierno de Navarra / Nafarroako Gobernua

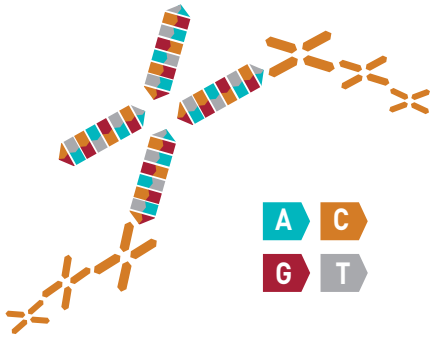
NS³ NAVARRE HEALTH

WHAT IS A GENOME?

Our genes store complete information with instructions on the functions and development of the body throughout our lifetime. Each set of instructions is called **a gene**. We have **about 20,000 genes in our body** and a significant number of them have been associated with different genetic disorders. Genes are stored in structures called **chromosomes**, which are like instruction manuals, whereas genes are like the chapters of those manuals.

The **genome** consists of all the genes in the body plus all other genetic information.

The genome is made up of **DNA**, which is a long sequence of more than three billion letters of four different kinds (A, C, T and G).



We now know that each letter can be important, even if it does not form part of a gene. This is the reason why we want to **analyze whole genomes**, which involves reading each and every letter. **The aim is to describe the cause, diagnosis and best treatment for as many patients as possible.**

Part of an individual's genome is unique to that person. However, most of our information is shared by other family members. Therefore, in some cases, it can be helpful to invite parents to participate in the study.

PROJECT GOALS

TO ASSESS GENOME SEQUENCING AS A CLINICAL TOOL

To assess the **clinical impact of genome sequencing** in children with a suspected genetic disorder.

To evaluate the use of studying the whole human genome as a tool for discovering the **genetic basis of autism spectrum disorder (ASD)** associated with comorbidity (simultaneous occurrence of other clinical manifestations).

TO IMPROVE PATIENT CARE

Some boys and girls with a suspected genetic disorder **whose specific cause has not been identified** can learn about the causes of their disorder, obtain a diagnosis for the first time and/or discover the treatment that would work best in their case.

TO ENCOURAGE RESEARCH

The data generated by this project can help the international scientific community **develop new areas of research** that generate knowledge and a better understanding of the causes of genetic disorders with the aim of **finding more effective methods of prevention and treatment**.

WHO CAN PARTICIPATE IN THE PROJECT AND HOW DOES THE ADMISSION PROCESS WORK?

NAGEN PEDIATRICS will allow genetic information to be collected from:

- **Patients with childhood cancer**
- **Hospitalized children with a suspected genetic disorder**
- **Patients with a positive newborn screening result: metabolic disorders**
- **Children with autism spectrum disorder**

Patients can be proposed by their medical specialist as candidates for participation, provided they meet specific selection criteria. If the family is interested, an appointment will be arranged with the patient and family members to provide them with detailed information on the project.
If the family decides to participate, they must sign an **informed consent** form, after which blood samples can be taken and anonymized before being sent for sequencing and analysis. This procedure ensures compliance with current legislation on personal data protection and confidentiality.

Data on the DNA in these samples will be stored and kept in servers that follow strict security protocols. The final report with genome results of clinical use will be included in each patient's medical record.