CURRICULUM VITAE

Guiomar Pérez de Nanclares Leal

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Birth place and data: 13-01-1973. Bilbao, Spain.

Education:

- **PhD in Biology,** University of the Basque Country, July 2002. "*Implication of the INS-VNTR region and the CTLA-4 gene in autoimmunity: analysis in type-1 diabetes, celiac disease and Addison disease*". Awarded with the highest mark "Sobresaliente Cum Laude"
- **BSc Biology**. University of the Basque Country, 1996
- **BSc Biochemistry**. University of the Basque Country, 1996.

Professional experience:

- **Main Researcher** January 2010-today. OSI Araba-Txagorritxu (Vitoria, Spain). Genetic analysis on rare diseases and Epigenetic studies of imprinting disorders.
- **Coordinator of the DNA Bank of the Hospital** (As part of the Basque Biobank for research www.biobancovasco.org). January 2010-today
- **Assistant Professor** September 2004-today. University of the Basque Country, Department of Biochemistry and Molecular Biology.
- **FIS (National Health System) Researcher** March 2004-December 2009 at Cruces University Hospital. Genetic and functional characterization of genes involved in monogenic diabetes and other endocrine disorders.
- **Postdoctoral Fellow**, 2002-2004. Cruces University Hospital (Barakaldo, Spain). Genetic diagnosis of endocrine diseases.

PhD works directed:

- Dr. JI San Pedro "*Heterogeneity of vitamin-D receptor polymorphisms in their susceptibility to autoimmunity: type 1 diabetes mellitus and celiac disease*".2006
- Dr. E.Fernández "*Clinic and genetic characterization of pseud hypoparathyroidism: analysis of GNAS locus*", 2009
- Dr. I. Garin "*Gene discovery on monogenic diabetes*", 2010
- Dr. A. Pereda"*Characterisation of patients with brachydactyly type E associated or not with hormonal resistance: clinical, genetic and radiological approach"*, 2015

Publications: Author and coauthor of 83 peer- review articles (h-index 20, WOS), with an accumulated impact factor higher than 320 and a citation average per article of 12.7. I have also published some articles in national journals, book chapters and, last year I have edited the book entitled "Guía de buenas prácticas clínicas en enfermedades de impronta, Clinical guidelines and best practice advice on imprinting disorders". My aim, reached during the years, is to publish, at least, 5 articles per year.

Summary of my research pathway: I started my research career in the area of genetic and immunological aspects of type 1 diabetes mellitus. In July 2002 I defended my PhD about a

series of genetic factors involved in polygenic diabetes. In 2004 I began a new line of research and moved from type 1 diabetes to monogenic diabetes. In 2007 I began to work independently on pseudohypoparathyroidism (PHP) and to establish international contacts related to this line. In January 2010 I joined Hospital Txagorritxu and initiated a new research group. I follow my research line on pseudohypoparathyroidism as I have become a National Reference for it and I have expanded it to some other imprinting disorders and epigenetic alterations associated to very specific types of cancer.
Our group is a national and international reference centre for the (epi)genetic study of PHP, which has given us the opportunity to join two international project: one, specific for PHP (EuroPHP net, supported by ESPE) and the other one related to imprinting disorders (Action COST BM1208). This international research networks has produced many collaborative papers with high internation impact.

PREVIOUS PROJECTS FUNDED

Projects funded as Principal Investigator

Gene discovery in monogenic diabetes
The main goal of this project was to identify new genes involved in the development of monogenic diabetes.
Funding body: Spanish National Ministry of Health FIS PI06/0690
Funding: 80.560€; Duration: 1/1/2007-31/12/2009

Pseudohypoparathyroidism and pseudopseudohypoparathyroidism: classification and clinic and genetic characterization
Goal: To analyze the complete GNAS locus in patients with pseudohypoparathyrodism in order to evaluate the genetic mechanisms involved in the disease
Funding body: Basque Department of Health (GV2008/11035) and BIOEF (BIO/ER/0001)
Funding: 37300€+39100€, respectively; Duration: 1/1/2009-31/12/2011

Genetic and environmental causes of congenital malformations
Goal: To analyze the putative association between assisted reproductive technique (ART) and chromosomal rearrangement or specific imprinting mutations in patients with congenital malformations
Funding body: Spanish National Ministry of Health ETES PI08/90503
Funding: 78.892€; Duration: 1/1/2009-31/12/2010

Hypomethylation syndrome characterization and its relation with assisted reproductive techniques
Objectives: The main objective of the project is to analyze the presence of hypomethylation syndrome in patients diagnosed with type PHP-Ib and its possible association with assisted reproductive techniques.
Funding body: Eugenio Rodríguez Pascual Fundation (FERP10/001)
Budget: 40.100€; Duration: 1/1/2010-31/12/2012

Molecular characterization of imprinting control region(s) for PHP: *cis* and *trans* approach
Objectives: (1) To identify PHP patients with imprinting defects; (2) To confirm the absence of pat20UPD or previously described deletions; (3) To characterize chromosome 20q for alterations in sequences putatively associated with imprinting defects; (4) To identify molecular defects at genes responsible for imprinting establishments.
Funding body: Spanish National Ministry of Health (PI10/00148)
Budget: 121.000€; Duration: 1/1/2011-31/12/2013

New genetic mechanisms involved in imprinting disorders

Objectives: (1) To generate standardized recording of phenotypes, to develop an imprinting disorders classification system and to develop clinical diagnostic and management guidelines and (2) To decipher the funtional (epi)genetic mechanisms underlaying imprinting disorders and to identify new imprinting disorders and new (epi)genetic causative factors.
Funding body: Spanish National Ministry of Health (PI13/00467)
Budget: 167.000€; Duration: 1/1/2014-31/12/2016

Developmental of a screening method to characterize differentially methylated regions involved in cancer an imprinting disorders
Main objective: to design, by two indenpendent technological strategies, kits for the characterization of the most common genetic and epigenetic alterations in imprinting disorders and some specific types of cancer.
Funding body: Health Department of Basque Government (GV2014/111017)
Budget: 62.272€; Duration: 1/10/2014-31/9/2017

Imprinting disorders: low grade mosaicism or new candidate genes?
The main objective of the present project is to develop an NGS-based technology to identify new underlying causes for imprinting disorders.
Funding body: Spanish National Ministry of Health (PI16/00073)
Budget: 167.000€; Duration: 1/1/2017-31/12/2019

Projects funded as Collaborator: In addition to the grants I received as PI, since the beginning of my career to 2012 I have participated in 9 additional funded projects as co-investigator.

Since 2011 I am also co-investigator in the project «***EuroPHPnet, a unique tool to decipher the Gsa dependent signalling pathways***». The objective is to build a structure that will facilitate the circulation of samples and thoughts in order to improve, the diagnosis and care of patients affected with pseudohypoparathyroidism or Albright Hereditary osteodystrophy. The project gathers of 7 investigators and funded has been awarded for meetings, circulation of samples and development of a core dataset.
Funding body: European Society for Paediatric Endocrinology
Funding for the Consortium: 60.000 €; Starting date: January 2012; Duration: 2 years

I am also member of the Management Committee for COST Action BM1208 on imprinting disorders

Signed: Guiomar Pérez de Nanclares
June 22, 2018