

Título del Proyecto	<b>Identificación y caracterización de mecanismos moleculares implicados en las Anomalías de la Diferenciación Sexual (ADS) mediante la aplicación de técnicas de secuenciación masiva y aCGH</b>
Nº de expediente asignado	PI15/01647
Abstract	<p>Disorders of sex development (DSD) are congenital conditions in which development of chromosomal, gonadal and genital sex is atypical. DSD are classified in three main diagnostic categories: DSD with sex chromosome abnormalities, 46,XY DSD (46,XY karyotype) and 46,XX DSD (46,XX karyotype). A large number of genes have been associated with the control of sexual differentiation (mainly genes coding for transcription factors, steroidogenesis enzymes, peptide hormones and receptors). Alteration in these genes is the molecular basis of a percentage of 46,XY DSD and 46,XX DSD patients. Likewise, there is a percentage of patients with DSD with no known molecular defect. The general objective of this project is the identification and characterization of the molecular basis of DSD in patients with unknown molecular diagnosis by implementing next generation sequencing (NGS) techniques and aCGH and to then transfer these results to routine genetic diagnosis. The specific objectives are: 1) Formation of a cohort of patients with DSD; 2) Design and validation of a next generation sequencing panel for genes involved in different</p>

	<p>pathways of sexual differentiation; 3) Mutation screening in patients with DSD and unknown genetic defect by the new NGS panel and aCGH, 4) Whole Exome Sequencing of a patient selection (familial trio) with unknown defect following the previous objective; 5) To functionally characterize the predicted pathogenic variants.</p>
Entidad Financiadora	Instituto de Salud Carlos III
Convocatoria:	Proyectos de Investigación en Salud – Conv. AES 2015
Importe de la ayuda	98.615€
Fechas de ejecución del proyecto	01/01/2016-31/12/2018
	Estas ayudas están financiadas por el Instituto de Salud Carlos III y cofinanciadas por FEDER, Programa Operativo Crecimiento Inteligente 2014-2020 “Una manera de hacer Europa”



Enlaces:

<http://www.ciberisciii.es/areas-tematicas/grupo-de-investigacion?id=17107>

<http://www.ciberisciii.es/Memorias/2016/FLIPS/Flip-CIBERER2017-cast/Memo-ER2016-Flip-CAST.html#p=11>

