Título del Proyecto	**PIK3CA Overgrowth Syndromes: Diagnosis, Phenotype and Clinical Guidelines**
Nº de expediente asignado	PIK3CA
Abstract	The presence of multiple clones of cells with various genotypes in the same individual is known as "somatic mosaicism", a pathogenic mechanism in which the mutations are not present in the germ-line but arise as a post-zygotic event, causing both cancer and highly variable developmental genetic syndromes. Recently, a group of segmental overgrowth syndromes caused by somatic mutations in the *PIK3CA* gene have been described. The group includes previously considered separate syndromes as Megalencephaly-Capillary malformation (MCAP) and CLOVES, along with some other variants as isolated adipose fibrodysplasia or hemimegalencephaly. However, as somatic mosaicism makes clinical expression variable in severity and location, the phenotypic spectrum of this group of segmental overgrowth syndromes is still to be elucidated, in order to prevent patients with unusual phenotypic manifestations to be excluded from the diagnosis. Another complication associated with the diagnosis of somatic mutations is that they can occur in low mosaics, making them difficult to detect by standard molecular techniques. This project will use Next Generation Sequencing (NGS) to perform an experimental and bioinformatic protocol, reliable and applicable to clinical practice, for the diagnosis of patients with *PIK3CA* segmental overgrowth syndromes, as a model for an NGS diagnostic panel to detect low mosaic mutations in developmental syndromes. As part of the clinical diagnostic process, we will evaluate those key clinical features that could suggest the diagnosis of a patient despite not meeting the strict diagnostic criteria established for MCAP and CLOVES. This will allow increasing the percentage of diagnosed patients. We will also develop clinical guidelines for both health professionals and patients and their families to give them access to this generally limited information, and we will promote the creation of a specific association of patients for these diseases.
Entidad Financiadora	Federación ASEM, FEDER, Fundación Isabel Gemio
Convocatoria:	Convocatoria “Todos somos raros, todos somos únicos” - 2014

Importe de la ayuda	87.815€
Fechas de ejecución del proyecto	15/02/2015 – 15/02/2017
	    
Enlaces:	https://www.ciberisciii.es/areas-tematicas/grupo-de-investigacion?id=17107