

SpainUDP Madrid, Spain

Brief description of the “Clinical Center” or “Services on undiagnosed rare diseases”

Research staff: Beatriz Martínez; Estrella López; Eva Bermejo; Francisco Javier Alonso; Isabel Cuesta; Sara Monzón; Gema Gómez; Julian Lara; Rosario Cazorla; Purificacion Ros; Gema Iglesias; Carlos Jimenez; Pablo Tutor; Enriqueta Roman; Susana Mellor; M.José Cabrejas. Existing program on undiagnosed rare diseases: SpainUDP is the Spanish Undiagnosed Rare Diseases Program, implemented by the Institute of Rare Disease Research, IIER, Instituto de Salud Carlos III -ISCIII. The ISCIII is the governmental organization for health research, acting also as a Funding Agency for Health Research at the National Health System and it is also a full member of IRDiRC. The ISCIII has signed an agreement with one of the main hospitals in Madrid and both act as a joint program. The SpainUDP website is <http://spainudp.isciii.es/home/>. From the official launch of SpainUDP in October 2015 to December 2018, 167 cases were accepted in SpainUDP and they were mainly paediatric. During this time period, 40 cases dropped out the program due to diverse reasons: they got their diagnosis independently of SpainUDP, they had a common disease instead of a rare disease, they decided to leave the program voluntarily, etc. The remaining 127 cases are distributed as follows: phenotypic and genotypic characterization was finished in 38 cases, of which 28 were diagnosed and in 7 cases we have identified candidate genes pending on functional studies and possibly related to new syndromes; 42 cases are pending on variants' validation by Sanger sequencing; in 12 cases, WES is ongoing; and the remaining 35 cases are being studied in depth for phenotypic characterization.

Past (six months) and future/upcoming events related to undiagnosed rare disease

Last 7th November 2018, in Madrid (Ministry of Science, Innovation and Universities) it was held the “Joint Japan-Spain Symposium on Medical Research. This symposium was an opportunity for the establishment of an official agreement between both countries (Japan and Spain) for supporting the collaboration between IRUD (Japanese RD Undiagnosed Program) and SpainUDP. In addition, SpainUDP and UDNI are always mentioned and described in all my talks around meetings in Spain. Some posters have been also presented in some national and European meetings.

Relevant scientific publications

Int J Environ Res Public Health. 2018;15(8). SpainUDP: The Spanish Undiagnosed Rare Diseases Program. López-Martín E, Martínez-Delgado B, Bermejo-Sánchez E, Alonso J; SpainUDP Network, Posada M.

Grants. a) grant award to announce funding for projects; b) Received grants from other organizations/institutions for undiagnosed rare diseases research.

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