Título del Proyecto	**Avances en la enfermedad de McArdle: Nuevas aproximaciones terapéuticas y desarrollo de un nuevo método diagnóstico no invasivo en pacientes.**
Nº de expediente asignado	PI13/00855
Abstract	McArdle disease is an autosomic recessive myopathy caused by mutations in the PYGM gene, which results in myophosphorylase deficiency. Patients typically experience `exercise intolerance', frequently accompanied by rhadbomyolysis, hyper-CK-emia and myoglobinuria. There is no curative therapy for the disease. The present project is basically focused in two different main goals: 1) evaluation of potential curative therapies of the disease using the following approaches: i) gene therapy designed to introduce the PYGM and PYGB isoforms in the skeletal muscle, ii) pharmacological treatment with a collection of read-through compounds (Ataluren, RTC13 and Amlexanox) that inhibit the effect of non-sense mutations in PYGM, iii) pharmacological treatment with an inhibitor of histone deacetylation (valproic acid) that would allow the reexpression of the hepatic and/or brain isoforms in the skeletal muscle; these approaches will be applied in both the McArdle mouse model generated by our group and also in skeletal muscle cell cultures from patients of the national registry and the murine model, representative of the principal mutations causing the disease. 2) development of a novel non-invasive diagnostic method for

	McArdle disease based on flow citometry, using blood from 100 patients of the national registry and from 100 sex and age-matched controls.
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Enlaces:	http://www.ciberisciii.es/areas-tematicas/grupo-de-investigacion?id=17109 http://www.ciberisciii.es/Memorias/2016/FLIPS/Flip-CIBERER2017-cast/files/basic-html/page11.html