IRCCS Istituto Giannina Gaslini
and
Università degli Studi di Genova

Manifestation of interest

Clinical outcome and quality of life in patients with ARPC1B deficiency managed conservatively or with hematopoietic stem cell transplantation

Project short name: ARPC1B_outcome

Brief rationale: ARPC1B deficiency results in a combined immunodeficiency characterized by early clinical onset, recurrent infections related to impaired T-cell function, allergic manifestations, and platelet abnormalities with bleeding tendency. Although most patients with ARPC1B mutations tolerate transplant conditioning, with a high rate of engraftment and resolution of immunodeficiency, there is currently a lack of studies comparing the clinical outcome and quality of life of patients undergoing transplantation or treated conservatively.

Objectives:
To compare ARPC1B patients managed conservatively and with HSCT assessing

1. Clinical outcome
2. Quality of life

What we need from you:
1. Fill out a Clinical Report Form with clinical and laboratory information of ARPC1B patients followed at your Centre.
2. Having the quality-of-life questionnaires filled in: PedsQL 4.0 and SDQ for children and SF-12 for adults respectively.

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