

Haematologica
HAEMATOL/2017/167601
Version 3

Mutations in the adaptor-binding domain and associated linker region of p110 β ; cause Activated PI3K β Syndrome 1 (APDS1)

Lucie Heurtier, Hicham Lamrini, Loïc Chentout, Marie-Céline Deau, Amine Bouafia, Jérémie Rosain, Jean-Marc Plaza, Mélanie Parisot, Benoit Dumont, Delphine Turpin, Etienne Merlin, Despina Moshous, Nathalie Aladjidi, Bénédicte Neven, Capucine Picard, Marina Cavazzana, Alain Fischer, Anne Durandy, Jean-Louis Stephan, and Sven Kracker

Disclosures: The work was supported by the "Institut National de la Santé et de la Recherche Médicale, the Fondation pour la Recherche Médicale (grant number: ING20130526624), la Ligue Contre le Cancer (Comité de Paris), the Centre de Référence Déficiés Immunitaires Héritaires (CEREDIH), the Agence Nationale de la Recherche (ANR-15-CE15-0020 [ANR-PIKimun]) and a government grant managed by the French Agence Nationale de la Recherche as part of the "Investments for the Future" program (ANR-10-IAHU-01). L.H. was supported by a grant from the Bettencourt-Schueler foundation through the Imagine MD-PhD program. S.K is a Centre National de la Recherche Scientifique (CNRS) researcher.

Contributions: SK and LH designed research; LH, HL, LC, MCD, AB, MP performed research; BD, JR, DM, DT, CP, NA, BN, EM and JLS provided patient care and collected data; SK, AD, AF, MC, JMP, LH analyzed and interpreted data; SK and LH wrote the manuscript and all authors commented on manuscript.