

Supplementary Table 1. Classification of patients with a suspicion of LRBA deficiency

Group	Criteria
Possible	<p>Patients who met the following criteria are considered as suspected of LRBA deficiency:</p> <p>I. Unknown genetic cause of their disease and any of the following diagnosis: IIa. ALPS-phenotype (Autoimmune lymphoproliferative syndrome of undetermined genetic cause), IIb. CVID (Common Variable Immunodeficiency), IIc. Child-onset hypogammaglobulinemia, IId: early-onset Inflammatory Bowel Disease, i.e. age of onset <10 years, and IIe: Autoimmune cytopenias of unknown etiology. III. Signed patient consent. IV. Availability of samples.</p>
Probable	<p>Possible LRBA-deficient patients with absence or reduced LRBA protein expression tested by flow cytometry or Western blot, or as suggested here, with a LRBA-MFI ratio below 2.6</p>
Definitive	<p>Probable LRBA-deficient patients with biallelic mutations in <i>LRBA</i></p>
Not LRBA-deficient	<p>Patients with a suspicion of LRBA deficiency but with wild type sequence of <i>LRBA</i></p>