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Pediatric-onset Evans syndrome: Heterogeneous presentation and high frequency of monogenic disorders including LRBA and CTLA4 mutations

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Titre	Pediatric-onset Evans syndrome: Heterogeneous presentation and high frequency of monogenic disorders including LRBA and CTLA4 mutations
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Auteur	Besnard, Caroline [1], Levy, Eva [2], Aladjidi, Nathalie [3], Stolzenberg, Marie-Claude [4], Magérus-Chatinet, Aude [5], Alibeu, Olivier [6], Nitschke, Patrick [7], Blanche, Stéphane [8], Hermine, Olivier [9], Jeziorski, Eric [10], Landman-Parker, Judith [11], Leverger, Guy [12], Mahlaoui, Nizar [13], Michel, Gérard [14], Pellier, Isabelle [15], Suarez, Felipe [16], Thuret, Isabelle [17], de Saint-Basile, Geneviève [18], Picard, Capucine [19], Fischer, Alain [20], Neven, Benedicte [21], Rieux-Laucat, Frédéric [22], Quartier, Pierre [23]
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Résumé en anglais	Evans syndrome (ES) is defined by the combination of autoimmune hemolytic anemia and immune thrombocytopenia. Clinical presentation includes manifestations of immune dysregulation, found in primary immune deficiencies, autoimmune lymphoproliferative syndrome with FAS (ALPS-FAS), Cytotoxic T Lymphocyte Antigen-4 (CTLA-4) and Lipopolysaccharide-Responsive vesicle trafficking Beige-like and Anchor protein (LRBA) defects. We report the clinical history and genetic results of 18 children with ES after excluding ALPS-FAS. Thirteen had organomegaly, five lymphocytic infiltration of non-lymphoid organs, nine hypogammaglobulinemia and fifteen anomalies in lymphocyte phenotyping. Seven patients had genetic defects: three CTLA4 mutations (c.151C>T; c.109+1092_568-512del; c.110-2A>G) identified by Sanger sequencing and four revealed by Next Generation Sequencing: LRBA (c.2450+1C>T), STAT3 gain-of-function (c.2147C>T; c.2144C>T) and KRAS (c.37G>T). No feature emerged to distinguish patients with or without genetic diagnosis. Our data on pediatric-onset ES should prompt physicians to perform extensive screening for mutations in the growing pool of genes involved in primary immune deficiencies with autoimmunity.
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Liens

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