Report of clinical presentations and two novel mutations in patients with Wiskott-Aldrich Syndrome/X-linked thrombocytopenia

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Abstract

Wiskott-Aldrich syndrome (WAS)/X-linked thrombocytopenia (XLT) is a rare X-linked disease characterized by thrombocytopenia, eczema, and recurrent infection. In addition, WAS/XLT increases incidence of autoimmune diseases and malignancies. We reported 7 male patients, 2 with WAS and 5 with XLT, from 6 different families. Two novel mutations, p.Gly387GlufsTer58 and p.Ala134Asp, were identified in patients with WAS. Both patients had severe clinical phenotypes compatible with classic WAS and developed lethal outcomes with intracranial hemorrhage. Other than that, one patient with XLT developed pineoblastoma.

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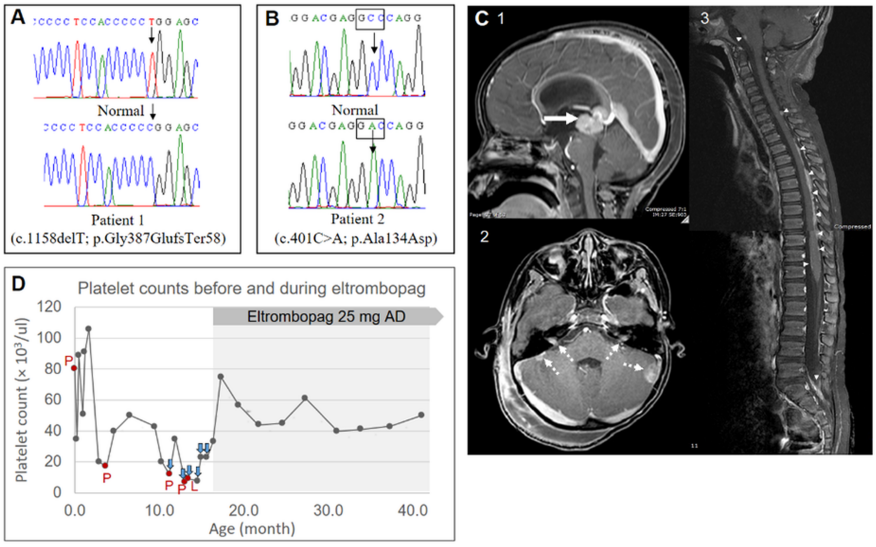

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TABLE 1.doc available at https://authorea.com/users/732070/articles/710685-report-of-clinical-presentations-and-two-novel-mutations-in-patients-with-wiskott-aldrich-syndrome-x-linked-thrombocytopenia