Natural history of GATA2 deficiency in a survey of 79 French and Belgian patients

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Résumé en anglais	Heterozygous germline mutations strongly predispose to leukemia, immunodeficiency, and/or lymphoedema. We describe a series of 79 patients (53 families) diagnosed since 2011, made up of all patients in France and Belgium, with a follow up of 2249 patients/years. Median age at first clinical symptoms was 18.6 years (range, 0-61 years). Severe infectious diseases (mycobacteria, fungus, and human papilloma virus) and hematologic malignancies were the most common first manifestations. The probability of remaining symptom-free was 8% at 40 years old. Among the 53 probands, 24 had missense mutations including 4 recurrent alleles, 21 had nonsense or frameshift mutations, 4 had a whole-gene deletion, 2 had splice defects, and 2 patients had complex mutations. There were significantly more cases of leukemia in patients with missense mutations (n=14 of 34) than in patients with nonsense or frameshift mutations (n=2 of 28). We also identify new features of the disease: acute lymphoblastic leukemia, juvenile myelomonocytic leukemia, fatal progressive multifocal leukoencephalopathy related to the JC virus, and immune/inflammatory diseases. A revised International Prognostic Scoring System (IPSS) score allowed a distinction to be made between a stable disease and hematologic transformation. Chemotherapy is of limited efficacy, and has a high toxicity with severe infectious complications. As the mortality rate is high in our cohort (up to 35% at the age of 40), hematopoietic stem cell transplantation (HSCT) remains the best choice of treatment to avoid severe infectious and/or hematologic complications. The timing of HSCT remains difficult to determine, but the earlier it is performed, the better the outcome.
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Liens

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