

GATA2 Deficiency

Early identification for improved clinical outcomes

Kristen Cole, DNP, RN, OCN®, BMTCN®, Daniele N. Avila, MSN, CRNP, ANP-BC, Mark Parta, MD, Bazetta Blacklock Schuver, BSN, BS, RN, Steven M. Holland, MD, Nirali Shah, MD, and Dennis Hickstein, MD

BACKGROUND: Patients with GATA2 deficiency present with nontuberculous mycobacterial infections, severe viral infections (particularly refractory human papillomavirus disease), lymphedema, myelodysplastic syndrome (MDS), and acute myeloid leukemia. Patients with GATA2 deficiency who undergo allogeneic hematopoietic stem cell transplantation prior to the development of life-threatening infections or cytogenetic abnormalities may have optimal clinical outcomes.

OBJECTIVES: The aim of this article is to determine ways in which oncology nurses can identify GATA2 deficiency in patients early and optimize treatment decisions.

METHODS: A case study is presented of a 33-year-old man with recurrent infections and MDS and his two sons, all of whom were found to have the same GATA2 mutation.

FINDINGS: Oncology nurses play an important role in early detection and identification by interviewing patients and obtaining a complete and thorough family history.

KEYWORDS

GATA2 deficiency; myelodysplastic syndrome; acute myeloid leukemia

DIGITAL OBJECT IDENTIFIER

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INDIVIDUALS WHO HAVE GATA2 DEFICIENCY experience severe, life-threatening infections, respiratory problems, deafness, lymphedema, and leukemia. As reviewed by Spinner et al. (2014), GATA2 deficiency was first described by four different research groups, resulting in the following four different names: monocytopenia and mycobacterial infections, or MonoMAC (Hsu et al., 2011; Vinh et al., 2010); deficiencies in dendritic, monocyte B, and natural killer lymphoid cells (Bigley et al., 2011; Dickinson et al., 2011); familial myelodysplastic syndrome (MDS)/acute myeloid leukemia (AML) (Hahn et al., 2011); and Emberger syndrome (Mansour et al., 2010; Ostergaard et al., 2011). Signs and symptoms of GATA2 deficiency first appear in childhood or early adulthood, but patients and family members can have variable presentations. The incidence of this disease is higher than originally thought; as many as 72% of adolescents with MDS and monosomy 7 (a single copy of chromosome 7) have an underlying GATA2 deficiency (Wlodarski et al., 2016).

Allogeneic hematopoietic stem cell transplantation (HSCT) is currently the only definitive therapy for GATA2 deficiency (Cuellar-Rodriguez et al., 2011; Dickinson et al., 2014; Donadieu et al., 2018; Spinner et al., 2014; Wlodarski et al., 2016). HSCT is a treatment option for patients with recurrent, treatment-refractory, severe infections (Cuellar-Rodriguez et al., 2011). Allogeneic HSCT replaces the defective immune and hematopoietic cells in a patient with stem cells from a healthy donor, thereby restoring normal immune and marrow function. However, HSCT has little impact on some manifestations, such as lymphedema. GATA2 deficiency is familial in about half of patients with the disease and de novo in the remainder (Spinner et al., 2014). The GATA2 mutation in the familial cases is inherited as Mendelian dominant, and it functions by loss of activity, or haploinsufficiency.

Healthcare providers can influence the clinical outcomes of these patients by obtaining a detailed family history. This is particularly important for patients with newly diagnosed MDS or AML for whom a pattern of inheritance and whether other family members are potentially affected need to be identified to explore the possibility of a GATA2 mutation. Patients with a GATA2 mutation who undergo HSCT prior to developing life-threatening infections or cytogenetic abnormalities have considerably better disease-free outcomes.

GATA2

The syndrome of monocytopenia and mycobacterial infections, typically *Mycobacterium avium*, termed MonoMAC, was first recognized at the National Institutes of Health (NIH) (Vinh et al., 2010). It was shown that