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AIRE	Probe	NM_000383.2
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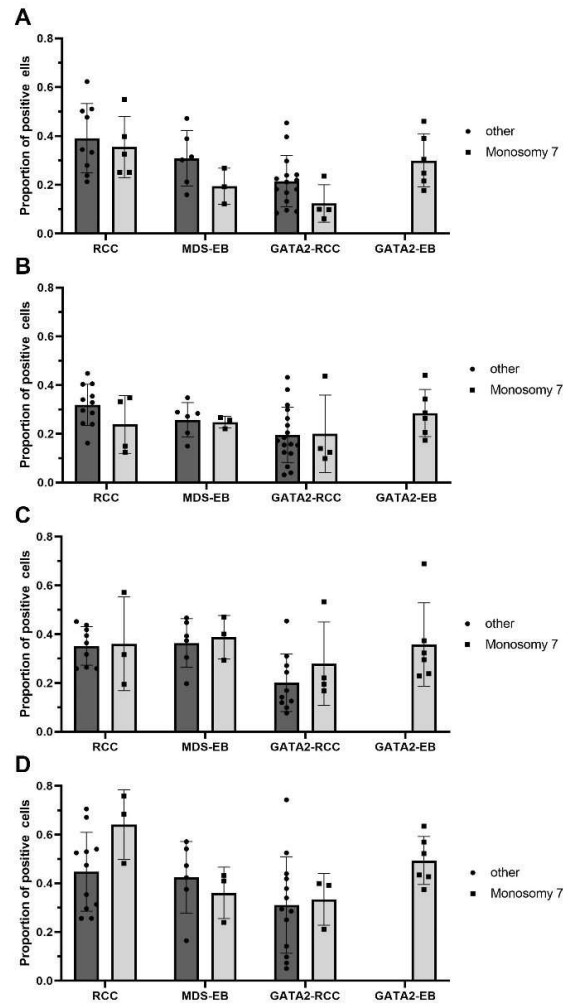
Supplementary Table S1: NanoString nCounter® Human PanCancer Immune Profiling Panel. Table of genes included in the gene expression profiling panel used in this study.

<i>GATA2</i> gene location	Genotype	
Exon 2	c.161C>A	p.S54X
Exon 3	c.303delC	p.A103Qfs*16
	c.341delA	p.N114Tfs*5
	c.351C>G	p.T117T
	c.416_417delCT	p.S139CfsTer45
	c.414_417delCTCT	p.S139CfsX78
	c.448G>T	p.G150*
	C599delG	p.G200VfsX18
	c.599dupG	p.S201*
Exon 4	c.981G>A	p.G327G (within N-ZF)
Intron 4-5	c.1018-10_1037del30bp	
Exon 5	c.1037_1046del	p.G346Vfs*38 (between C-ZF and N-ZF)
	c.1045T>A	p.C349S (within C-ZF)
	c.1061C>T	p.T354M (within C-ZF)
	c.1084C>T	p.R362* (within C-ZF)
	c.1084C>T	p.R362fsx* (within C-ZF)
	c.1109G>T	p.C370F (within C-ZF)
	c.1113C>A	p.N371K (within C-ZF)
	c.1128C>G	p.Y376X (within C-ZF)
Intron 5-6	c.1143+1G>A	
Exon 6	c.1186C>T	p.R396W (within C-ZF)
	c.1187G>A	p.R396Q (within C-ZF)
	c.1215G>T	p.K405N

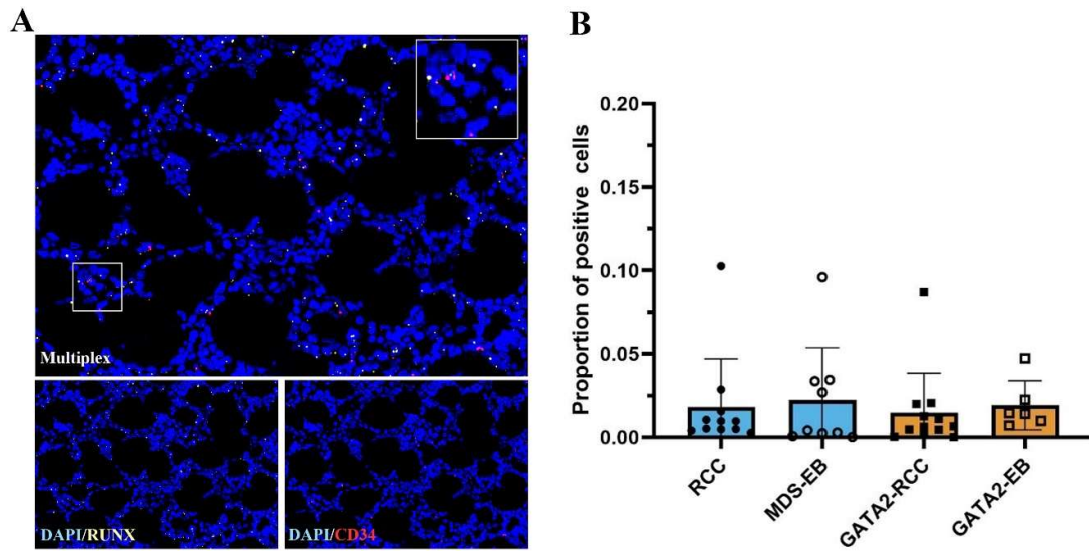
Supplementary Table S2: Distinct mutations found in pediatric patients with a *GATA2* deficiency and their localization on the *GATA2* gene. Table of 23 distinct mutations identified in n=25 patients with *GATA2* deficiency. Genotype unknown for n=5 patients. C-ZF: C-zinc finger; N-ZF: N-zinc finger.

Differentially expressed gene in GATA2-RCC versus GATA2-EB	Log2 fold change GATA2-RCC versus GATA2-EB	Std error (log2) GATA2-RCC versus GATA2-EB
<i>BCL2</i>	-1.200	0.294
<i>BMI1</i>	-1.400	0.312
<i>CCL18</i>	-2.140	0.232
<i>CCRL2</i>	1.220	0.281
<i>CD34</i>	-3.420	0.540
<i>CXCR1</i>	1.810	0.283
<i>IFNL1</i>	0.876	0.179
<i>IL18RAP</i>	2.860	0.480
<i>LCN2</i>	2.680	0.464
<i>LGALS3</i>	2.100	0.363
<i>MEF2C</i>	-1.760	0.430
<i>OSM</i>	1.230	0.250
<i>REPS1</i>	-1.310	0.296
<i>TRFC</i>	2.000	0.497
<i>TXK</i>	-1.240	0.278

Supplementary Table S3: Gene expression data for the 15 most differentially expressed genes in patients with a GATA2 deficiency in early and advanced disease stage in alphabetical order. GATA2-RCC: refractory cytopenia of childhood with additional *GATA2* mutation; GATA2-EB: MDS with excess blasts with additional *GATA2* mutation.



Supplementary Figure S1: Impact of the karyotype of patients on the expression of GATA2 target genes. *In situ* hybridization of GATA2 target genes *RUNX1*, *EZH2*, *IKZF1*, and *LYL1* for patients diagnosed with refractory cytopenia of childhood (RCC) or myelodysplastic syndrome with excess blasts (MDS-EB) and patients with RCC or MDS-EB and additional *GATA2* mutation (GATA2-RCC, GATA2-EB). Quantification of *RUNX1*- (A), *EZH2*- (B), *IKZF1*- (C), and *LYL1*-positive cells (D) revealed no statistically significant differences for patients with monosomy 7 or other karyotypes (normal karyotype, trisomy 8 and other cytogenetic aberrations) among pediatric patients. RCC: Monosomy 7 n=5, other n=12; MDS-EB: Monosomy 7 n=3, other n=7; GATA2-RCC: Monosomy 7 n=5, other n=16; GATA2-EB: Monosomy 7 n=6, other n=0.



Supplementary Figure S2: Comparable expression of *RUNX1*-positive hematopoietic progenitors in all patient groups, regardless of *GATA2* mutational status and disease stage. Multiplex *in situ* hybridization (**A**) of *RUNX1* (yellow) and *CD34* (red) with DAPI counterstain (blue). Quantification of *RUNX1*-positive hematopoietic progenitors (**B**) does not vary across all four patient groups, regardless of *GATA2* mutational status or disease progression.