Supplementary Table 1. Common variants found in the screening of *GATA2*, *GATA4* and *GATA6* genes.

Gene	Position	dbSNP ID	Nucleotide variant	Amino acid change	MAF (%)		p	MAF (%)	
					Patients	Controls		ESP	1000G
GATA2	I1	rs559062253	c.-46+13C>T	-	T=0.9	T=0.8	1.00	-	-
	E2	rs1573858	c.15C>G	p.Pro5=	C=26.2	C=28.5	0.43	C=34.9	C=36
	E3	rs2335052	c.490G>A	p.Ala164Thr	A=15.3	A=14.3	0.65	A=16.2	A=17.8
		rs34870876	c.564G>A	p.Thr188=	C=3.8	C=5.2	0.36	C=5.3	C=6.6
	I4	rs11708606	c.1018-19C>T	-	T=26.2	T=28.7	0.39	T=19	T=20.2
	I5	rs2713604	c.1143+203A>G	-	A=30	A=27.3	0.36	-	A=31.4
	E6	rs34172218	c.1233G>A	p.Ala411=	A=3.1	A=4.8	0.20	A=2.9	A=2.4
		rs3803	c.*482C>T	-	T=23.6	T=28	0.13	-	T=20.3
		rs45463801	c.*546C>T	-	T=6.6	T=6.7	1.00	-	T=4.1
GATA4	E1	rs61277615	c.-534C>T	-	T=3.3	T=4.7	0.33	-	T=8.2
	I2	rs10503425	c.617-64G>C	-	C=10.6	C=10	0.75	-	C=14.8
	E6	rs3729856	c.1129A>G	p.Ser377Gly	G=9	G=9.2	1.00	G=14	G=12.9
GATA6	I6	rs3764962	c.1620+7A>G	-	G=1.4	G=2.7	0.19	G=1.3	G=1.8

E: exon; I: intron; dbSNP ID: single nucleotide polymorphism identification in the database dbSNP; MAF: minor allele frequency; p: p-value; ESP:Exome Sequencing Project in European American population;1000G: 1000 Genomes Project in European populations. Statistical significance was considered as p-value <0.002 (0.05/22).Positions of variants based on GATA2 RefSeq NM_032638.4, NP_116027.2;GATA4 RefSeq NM_002052.3, NP_002043.2; GATA6 RefSeq NM_005257.3, NP_005248.2.

Supplementary Table 2.Summary of *in silico* analyses of rare variants found in *GATA2*, *GATA4* and *GATA6* genes.

Gene	Nucleotidevariant	Amino acidchange	Mutationtaster	SIFT	Polyphen-2	Other *in silico* tools
GATA2	c.-77G>A	-	Disease Causing	-	-	Alterations on mRNA secondary structure. Affect binding sites for transcription factors.
	c.*508G>A	-	Polymorphism	-	-	No effect.
GATA4	c.-306C>T	-	Polymorphism	-	-	No effect.
	c.-294G>T	-	Polymorphism	-	-	Alterations on mRNA secondary structure.
	c.699G>A	p.Thr233=	Disease Causing	-	-	No effect.
	c.783+51G>A	-	Polymorphism	-	-	-
	c.822C>T	p.Cys274=	Disease Causing	-	-	No effect.
	c.909+25G>A	-	Polymorphism	-	-	-
	c.1027G>A	p.Ala343Thr	Polymorphism	Tolerated	Benign	Aminoacid conserved across mammals. Alterations on mRNA secondary structure. Changes in protein phosphorylation.
	c.1056C>T	p.Asn352=	Polymorphism	-		No effect.
GATA6	c.-181G>A	-	Disease Causing	-		No effect.
	c.296T>C	p.Val99Ala	Polymorphism	Damaging	Benign	No effect.
	c.849G>A	p.Ala283=	Polymorphism	-	-	No effect.
	c.851C>G	p.Ala284Gly	Polymorphism	Tolerated	Damaging	No effect.
	c.970_978del	p.His324_His326del	Polymorphism	Damaging	-	No effect.
	c.1302+20C>T	-	Polymorphism	-	-	-
	c.1374C>T	p.Asn458=	Disease Causing	-	-	No effect.
	c.1663C>G	p.Pro555Ala	Disease Causing	Tolerated	Damaging	Aminoacid conserved across mammals. Alterations on mRNA secondary structure.

-: not analyzed. Positions relative toGATA2RefSeq NM_032638.4, NP_116027.2; GATA4RefSeqNM_002052.3, NP_002043.2; GATA6RefSeq NM_005257.3, NP_005248.2.