GATA6 Mutation	Protein change	Mutation type	N carriers (N probands)	Pancreatic features	Cardiac features	Other features	Inheritance	References
c.606_609dup	p.(His204fs)	Frameshift	1(1)	PD NDM EPI	PTA PDA	Gallbladder agenesis Anomalous hepatic Blood flow and patent ductus venous Hydronephrosis/-ureter	ND	(Stanescu, Hughes, Patel, & De León, 2015)
c.635_660del	p.(Pro212fs)	Frameshift	4(1)	PD NDM EPI	PDA PS TOF	Gallbladder agenesis CDH Intestinal malrotation	Inherited(maternal)	(Yau et al. 2017)
c.701del	p.(Pro234fs)	Frameshift	1(1)	PD NDM EPI	ASD VSD	None	de novo	(Allen et al.2011)
c.705C > G	P.(Tyr235Ter)	Nonsense	1(1)	PD CDM	ASD VSD PDA	Scoliosis	de novo	(Sanchez-Lechuga et al. 2020)
c744del	p.(Pro249fs)	Frameshift	1(1)	PD NDM EPI	PTA HPA ASD VSD	Gallbladder agenesis PLE Developmental delay Inguinal hernia Congenital hip dysplasia	de novo	(McMillan, Girgis, & Sellers, 2016)
c.754_905del	p.(Ala252fs)	Frameshift	1(1)	PD NDM EPI	TOF	None	de novo	(Gong et al., 2013)
c.877_880delinsTAC	p.(Val293fs)	Frameshift	1(1)	PD NDM EPI	TOF	Biliary atresia Microcephaly Learning difficulties Inguinal hernia	ND	(Allen et al., 2011)
c.899_902dup	p.(Ala302fs)	Frameshift	2(1)	PD NDM EPI	DORV VSD PS	Gallbladder agenesis Paucity of intrahepatic bile ducts Secondary hypothyroidism Bilateral posterior embryotoxon	Inherited(maternal)	(Ferreira, Devadason, Denvir, Seale,& Gupte, 2017)
c.951_954dup	p.(Leu319fs)	Frameshift	1(1)	PD NDM EPI	PTA ASD VSD TI TGA	Gallbladder agenesis Partial biliary atresia Symmestric cortical atrophy	ND	(Gong et al., 2013)
c.964_970del	p.(Tyr323fs)	Frameshift	1(1)	PD NDM EPI	MS PDA	Gallbladder agenesis Biliary atresia	de novo	(Chao et al., 2015)
c.968dup	p.(Tyr323*fs)	Frameshift	1(1)	PD NDM EPI	VSD ASD PS	Abnormal mesenteric veins	de novo	(Eifes et al., 2013)
c.969C>A	p.(Tyr323*)	Nonsense	2(1)	PD NDM ADM EPI	ASD PDA	CDH	Inherited(parnal)	(De Franco et al., 2013)
c.1036_1042del	p.(Thr346fs)	Frameshift	2(1)	PD NDM ADM EPI	TOF	Hypothyroidism Learning difficulties	Inherited(parnal)	(De Franco et al., 2013)
c.1108_1121dup	p.(Gly375fs)	Frameshift	1(1)	PD NDM EPI	TOF	None	de novo	(Allen et al., 2011)
c.1136-2A>G		Splicing	2(1)	PD NDM CDM EPI	PDA	Hepatic dysfunction	Inherited(parnal)	(De Franco et al., 2013)
c.1242C>A	p.(Cys414*)	Nonsense	2(2)	PD NDM ADM EPI	ASD ASD PS PDA TGA Tricuspid atresia	Renal dysfunction (persistent proteinuria)	de novo*2	(Tuhan et al., 2015 & Michelle L. Miles et al.,2020)
c.1291C > T	p.(Gln431*)	Nonsense	2(1)	PD NDM ADM	ASD VSD PDA PS	Gallbladder agenesis CDH Scoliosis	Inherited(maternal)	(Odile Gaisl et al.2019)
c.1303-10C>G		Splicing	1(1)	PD NDM EPI	IAA	Gallbladder agenesis	de novo	(Allen et al., 2011)

c.1303-1G>T		Splicing	2(1)	NDM EPI	ASD PS	Developmental delay Hypothyroidism	Inherited(maternal)	(De Franco et al., 2013)
c.1330T>C	p.Cys444Arg	Missense	3(1)	PD NDM ADM EPI	DORV PS PDA VSD ASD PTA	Hypoganadism Growth hormone deficiency Umbilical hernia Bifid left pelvicalyceal system Right hydrocele Right epididymal cyst Proteinuria Dysmorphic features Mild-moderate Neurocognitive impairment Hypospadias	Inherited(paternal)	(Yang Timothy Du, 2020)
c.1339T>C	p.(Cys447Arg)	Missense	1(1)	NDM	None	Hypothyroidism	ND	(De Franco et al., 2013)
c.1354A>G	p.(Thr452Ala)	Missense	1(1)	PD NDM EPI	ASD	Developmental delay Intestinal perforation	de novo	(Allen et al., 2011)
c.1366C>T	p.(Arg456Cys)	Missense	4(4)	PD NDM CDM EPI	PTA VSD TOF IAA	Umbilical hernia Developmental delay Seizures Hypothyroidism Absent gall bladder Thrombocytopenia Neonatal stroke Adrenal insufficiency	de novo*4	(Nikhil Raghuram et al.202,Allen et al.2011,Sanyoura et al.2018)
c.1367G>A	p.(Arg456His	Missense	2(2)	PD NDM ADM EPI	PDA VSD HLPA	Developmental delay Gallbladder agenesis Renal duplex collecting system	de novo*2	(Allenby al,201,Doris Škoric-Milosavljevet al.2019)
c.1396A>G	p.(Asn466Asp)	Missense	1(1)	PD NDM EPI	PDA	Gallbladder agenesis Intestinal malrotation Microcolon Developmental delay Epilepsy Transient hypothyroidism	ND	(Allen et al, 2011)
c.1397A>G	p.(Asn466Ser)	Missense	1(1)	PD NDM	ASD PS PDA	Transient idiopathic neonatal cholestasis Hypoglycemia episodes Low growth and cortisol level	de novo	(Catli et al., 2013)
c.1399G>A	p.(Ala467Thr)	Missense	1(1)	PD NDM EPI	ASD PS	Pituitary agenesis Learning difficulties Seizures	ND	(Allen et al., 2011)
c.1406G>A	p.(Gly469Glu)	Missense	2(1)	PD NDM ADM EPI	None	Hepatomegaly Developmental delay Hemiplegia Hypothyroidism	Inherited(paternal)	(De Franco et al., 2013)
c.1417A>C	p.(Lys473Gln)	Missense	1(1)	PD NDM EPI	ASD	Gallbladder agenesis	de novo	(Allen et al., 2011)
c.1428+1G>T		Splicing	1(1)	PD NDM	ASD PDA	Developmental delay	de novo	(Chao et al., 2015)

				EPI		Anaemia		
c.1429-41_1441del		Splicing	1(1)	PD NDM EPI	PDA	None	de novo	(De Franco et al., 2013)
c.1429-8T>G		Splicing	1(1)	PD NDM EPI	Dextrocardia APW AVSD	Early cognitive and motor delay CDH	de novo	(De Franco et al., 2013)
c.1435A>G	p.(Arg479Gly)	Missense	1(1)	NDM ADM EPI	TGA VSD PDA	Cryptorchidism PLE	de novo	(De Franco et al., 2013,Doris Škori_x0019_c-Milosavljev et al.2019)
c.1448_1455del	p.(Met483fs)	Frameshift	1(1)	PD NDM EPI	ASD VSD PS PDA HRV HTV	Gallbladder agensis	de novo	(Allen et al., 2011)
c.1477C>T	p.(Arg493*)	Nonsense	1(1)	PD NDM EPI	VSD PDA	CDH	de novo	(Suzuki et al., 2014)
c.1498_1501del	p.(Lys500fs)	Frameshift	1(1)	PD NDM EPI	DOLV VSD HPA PS PFO PDA	None	de novo	(Allen et al., 2011)
c.1504_1505del	p.(Lys502fs), p.(K562Dfs)	Frameshift	4(2)	PD NDM CDM EPI	VSD TOF PDA	Gallbladder agenesis Bicornuate uterus Neonatal pancytopenia Hypercativity	1*Inherited(maternal) 1*ND	(Bonnefond et al., 2012.Yorifuji et al., 2012)
c.1516+1G>C		Splicing	1(1)	PD NDM EPI	ASD PFO	transient hypothyroidism	de novo	(Allen et al., 2011)
c.1516+4A>G		Splicing	1(1)	PD NDM EPI	None	CDH	de novo	(Allen et al., 2011)

Table S. Overview of all described GATA6 mutations with pancreatic dysfunction and/or developmental defect.

List of abbreviations (alphabetically ordered):

ADM: Adult-onset Diabetes Mellitus
ASD: Atrial Septal Defect
APW: Aortopulmonary Window
AVSD: Atrioventricular Septal Defect
CDH: Congenital Diaphragmatic Hernia
CDM: Children-onset Diabetes Mellitus
DOLV: Double Outlet Left Ventricle
DORV: Double-outlet right ventricle
EPI: Exocrine Pancreatic Insufficiency
HLPA: Hypoplastic Left Pulmonary Artery
HPA: Hypoplastic Pulmonary Artery
HRV: Hypoplastic Right Ventricle
HTV: Hypoplastic Tricuspid Valve
IAA: Interrupted Aortic Arch
MS: Mitral valve Stenosis
NDM: Neonatal Diabetes Mellitus
PD: Pancreatic Dysplasia
PDA: Patent Ductus Arteriosus
PFO: Patent Forum Ovale
PLE: Protein Losing Enteropathy
PS: Pulmonary Stenosis
PTA: Persistent truncus arteriosus
TGA: Transposition of the Great Arteries
TI: Tricuspid valve Insufficiency
TOF: Tetralogy Of Fallot
VSD: Ventricular Septal Defect