Supplementary

Table S1 Clinical data of neonates with gene abnormality

Case	Sex	Gestational age	Birth weight	Family history	Arrhythmia	Structure	Heart failure	Treatment	Death
1	M	37	2370	Y	Ventricular fibrillation	-	-	Y	Y
2	M	37	2685	Y	Ventricular fibrillation	-	-	Y	Y
3	M	37.3	3300	Y	Supraventricular tachycardia	VSD	-	Y	-
4	M	41.3	3250	-	Junctional premature contraction	-	-	-	Y
5	M	40.9	3600	-	Supraventricular tachycardia	-	-	Y	-
6	M	35.7	3060	-	Ventricular tachycardia	-	Y	-	-
7	F	39.1	3540	-	Ventricular fibrillation	-	Y	Y	Y
8	F	35.5	2410	Y	Ventricular tachycardia	-	-	Y	-
9	F	40	3000	-	Sinus bradycardia	-	-	-	-
10	F	39.7	3700	-	Atrial premature contraction	-	-	-	-
11	F	37.5	3750	-	Ventricular premature contraction	-	-	-	-
12	M	39.3	3080	-	Junctional premature contraction	-	-	-	-
13	M	38.3	2800	Y	Grade I atrioventricular block	Hypertrophic cardiomyopathy	Y	Y	Y
14	M	29.3	1630	-	Grade I atrioventricular block	VSD, hypertrophic obstructive cardiomyopathy	Y	Y	Y
15	M	33.4	2220	-	Grade I atrioventricular block	VSD	Y	Y	-
16	F	38.9	3850	-	Right bundle branch block	Hypertrophic cardiomyopathy	-	-	-
17	F	37	2000	-	Supraventricular tachycardia	-	-	Y	-
18	M	37.3	2730	-	Long-QT syndrome	-	-	Y	-
19	M	36.5	2650	-	Sinus bradycardia	-	-	-	Y
20	F	29.3	1700	-	Grade I atrioventricular block	Hypertrophic cardiomyopathy, VSD	Y	Y	-
21	M	37.4	3000	-	Ventricular premature contraction	-	-	-	-
22	M	32.4	1500	-	Grade II atrioventricular block	VSD	-	-	-

VSD, ventricular septal defect.

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Table S2 Clinical symptom of neonates with gene abnormality

Case	Clinical symptom
1	Carnitine palmityl transferase II deficiency (cardiac shock, pulmonary hemorrhage, liver failure, renal failure, hyperammonemia, hyperkalemia, hypoglycemia, hypocalcemia)
2	Carnitine palmityl transferase II deficiency (severe anemia, hyperpotassemia, hypoglycemia, hypocalcemia)
3	Central hypoventilation syndrome, brain injury
4	Central hypoventilation syndrome
5	-
6	-
7	Carnitine palmityl transferase II deficiency (cardiac shock, liver failure, renal failure, convulsion, hyperammonemia, hyperkalemia, hypocalcemia, upper gastrointestinal bleeding)
8	Preterm, sepsis, anemia
9	Dysmorphic feature (auricle malformation, soft and hard cleft palate, small jaw, small eye spacing, bulging eyes), congenital esophageal atresia
10	Tuberous sclerosis (atrial and intracranial mass)
11	Grade I laryngeal cleft, Grade II subglottic stenosis
12	Inflammatory bowel disease
13	Carnitine palmityl transferase II deficiency (cardiac shock, liver failure, renal failure, hyperammonemia)
14	Hypertrophic obstructive cardiomyopathy, primary pulmonary hypertension, heart failure, hearing loss
15	Heart failure, preterm infant, bronchopulmonary dysplasia, congenital heart disease (ventricular septal defect, atrial septal defect, patent ductus arteriosus, pulmonary stenosis), Helicobacter hypertrophy, right repetitive kidney and hydronephrosis, cryptorchid, respiratory failure, feeding problem
16	Refractory epilepsy, encephalopathy, right ventricular hypertrophy, PDA, PFO, pulmonary hypertension, moderate left ear hearing abnormality, incomplete right bundle branch block
17	Recurrent supraventricular tachycardia, small for gestational age
18	Hypospadia
19	Multiple brain injuries, central hypoventilation syndrome
20	Cholestasis, congenital intestinal atresia
21	Dysmorphic feature, asphyxia, abnormal hearing, sepsis, pneumonia
22	Dysmorphic feature, congenital heart disease (aortic valve construction, ventricular septal defect, patent ductus arteriosus), severe asphyxia, sepsis, respiratory failure

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Table S3 Genetic data of neonates with gene abnormality

Case	Gene	Chr	Start position	Variant	Zygosity	Diseases	Inherit	PMID	Sanger	Classification
1	SLC25A20	3	48921485	NM_000387: exon3: c.270_271delinsT (p.F91Lfs*38)	Het	Carnitine-acylcarnitine translocase deficiency, [MIM:212138]; Carnitine-acylcarnitine translocase deficiency, [MIM:212140]	AR	11592821	Maternal	Pathogenic
		3	48921567	NM_000387: exon3: c.199-10T>G	Het	Carnitine-acylcarnitine translocase deficiency, [MIM:212138]; Carnitine-acylcarnitine translocase deficiency, [MIM:212141]	AR	10697964	Paternal	Pathogenic
2	SLC25A20	3	48921567	NM_000387: exon3: c.199-10T>G	Hom	Carnitine-acylcarnitine translocase deficiency, [MIM:212138]; Carnitine-acylcarnitine translocase deficiency, [MIM:212139]	AR	10697964	Paternal/ Maternal	Pathogenic
3	KCNQ1	11	2549192	NM_000218: exon2:c.421G>A (p.V141M)	Het	Long QT syndrome 1, [MIM:192500]; Jervell and Lange-Nielsen syndrome, [MIM:220400]; Short QT syndrome 2, [MIM:609621]; Atrial fibrillation, familial, 3, [MIM:607554]	AD/ AR	16109388	NA	Likely pathogenic
4	PHOX2B	4	41747992	NM_003924: exon3: c.756_776dup21(GCN)27	Het	Neuroblastoma with Hirschsprung disease, [MIM:613013]; Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, [MIM:209880]	AD	23231723	NA	Pathogenic
5	SCN5A	3	38592173	NM_198056: exon28: c.5690G>A (p.R1897Q)	Het	Atrial fibrillation, familial, 10, [MIM:614022]; Sick sinus syndrome 1, [MIM:608567]; Ventricular fibrillation, familial, 1, [MIM:603829]; Long QT syndrome 3, [MIM:603830]; Heart block, nonprogressive, [MIM:113900]; Cardiomyopathy, dilated, 1E, [MIM:601154]; Brugada syndrome 1, [MIM:601144]; Heart block, progressive, type IA, [MIM:113900]	AD/ AR	-	NA	Vus
6	DSG2	18	29100797	NM_001943: exon4: c.248T>G (p.L183R)	Het	Arrhythmogenic right ventricular dysplasia 10,[MIM:610193]; Cardiomyopathy, dilated, 1BB,[MIM:612877]	AD		De novo	Likely pathogenic
7	CPT2	1	53676110	NM_000098: exon4: c.764A>G (p.D255G)	Het	CPT II deficiency, myopathic, stress-induced, [MIM:255110]; CPT II deficiency, infantile, [MIM:600649]; CPT II deficiency, lethal neonatal, [MIM:608836]; CPT II deficiency, myopathic, stress-induced, [MIM:255110]; CPT II deficiency, infantile, [MIM:600649]; CPT II deficiency, lethal neonatal, [MIM:608836]	AD/ AR	-	Paternal	Likely pathogenic
		1	53676379	NM_000098: exon4: c.1033G>A (p.G345R)	Het	CPT II deficiency, myopathic, stress-induced, [MIM:255110]; CPT II deficiency, infantile, [MIM:600649]; CPT II deficiency, lethal neonatal, [MIM:608836]; CPT II deficiency, myopathic, stress-induced, [MIM:255110]; CPT II deficiency, infantile, [MIM:600649]; CPT II deficiency, lethal neonatal, [MIM:608837]	AD/ AR	-	Maternal	Likely pathogenic
8	KCNH2	7	150654411	NM_000238: exon5: c.1096C>T (p.R366X)	Het	Long QT syndrome 2, [MIM:613688]; Short QT syndrome 1, [MIM:609620]	AD/ AR	11468227	NA	Pathogenic
9	EFTUD2	17	42953301	NM_004247: exon10: c.869+1G>A	Het	Mandibulofacial dysostosis, Guion-Almeida type, [MIM:610536]	AD	-	NA	Likely pathogenic
10	TSC2	16	2131745	NM_000548: exon31: c.3760_3761delinsT (p.S1254Wfs*71)	Het	Tuberous sclerosis-2, [MIM:613254]	AD	-	NA	Likely pathogenic

Table S3 (*continued*)

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Table S3 (*continued*)

Case	Gene	Chr	Start position	Variant	Zygosity	Diseases	Inherit	PMID	Sanger	Classification
11	PTPN11	12	112926890	NM_002834: exon13: c.1510A>G (p.M504V)	Het	LEOPARD syndrome 1, [MIM:151100]; Metachondromatosis, [MIM:156250]; Noonan syndrome 1, [MIM:163950]	AD	11704759	De novo	Pathogenic
12	IL10RA	11	117860269	NM_001558: exon3: c.301C>T (p.R101W)	Het	Inflammatory bowel disease 28, early onset, autosomal recessive, [MIM:613148];Inflammatory bowel disease 28, early onset, autosomal recessive, [MIM:613148]	AR	22476154	Paternal	Pathogenic
		11	117864125	NM_001558: exon4: c.537G>A (p.T179T)	Het	Inflammatory bowel disease 28, early onset, autosomal recessive, [MIM:613148];Inflammatory bowel disease 28, early onset, autosomal recessive, [MIM:613149]	AR	29140941	Maternal	Pathogenic
13	SLC25A20	3	48897046	NM_000387: exon6: c.550G>T (p.G184X)	Het	Carnitine-acylcarnitine translocase deficiency, [MIM:212138]; Carnitine-acylcarnitine translocase deficiency, [MIM:212138]	AR	-	Maternal	Pathogenic
		3	48921567	NM_000387: exon3: c.199-10T>G	Het	Carnitine-acylcarnitine translocase deficiency, [MIM:212138]; Carnitine-acylcarnitine translocase deficiency, [MIM:212139]	AR	10697964	Paternal	Pathogenic
14	PTPN11	12	112926908	NM_002834: exon13: c.1528C>G (p.Q510E)	Het	LEOPARD syndrome 1, [MIM:151100]; Metachondromatosis, [MIM:156250]; Noonan syndrome 1, [MIM:163950]	AD	15948193	NA	Pathogenic
15	PTPN11	12	112888166	NM_002834: exon3: c.182A>G (p.D61G)	Het	LEOPARD syndrome 1, [MIM:151100]; Metachondromatosis, [MIM:156250]; Noonan syndrome 1, [MIM:163950]	AD	1704759	NA	Pathogenic
16	SCN2A	2	166170523	NM_021007: exon10: c.1288G>A (p.E430K)	Het	Epileptic encephalopathy, early infantile, 11, [MIM:613721]; Seizures, benign familial infantile, 3, [MIM:607745]	AD	-	De novo	Pathogenic
17	PKP2	12	33049541	NM_004572: exon1: c.125G>A (p.G42E)	Het	Arrhythmogenic right ventricular dysplasia 9, [MIM:609040]	AD	-	NA	Vus
18	SCN5A	3	38627487	NM_198056: exon16: c.2482C>G (p.L828V)	Het	Atrial fibrillation, familial, 10, [MIM:614022]; Sick sinus syndrome 1, [MIM:608567]; Ventricular fibrillation, familial, 1, [MIM:603829]; Long QT syndrome 3, [MIM:603830]; Heart block, nonprogressive, [MIM:113900]; Cardiomyopathy, dilated, 1E, [MIM:601154]; Brugada syndrome 1, [MIM:601144]; Heart block, progressive, type IA, [MIM:113900]	AD/ AR	23631430	De novo	Pathogenic
19	PHOX2B	4	41748007	NM_003924: exon3: c.756_776dup21(GCN)27	Het	Neuroblastoma with Hirschsprung disease, [MIM:613013]; Central hypoventilation syndrome, congenital, with or without Hirschsprung disease, [MIM:209880]	AD	23231723	NA	Pathogenic
	SCN5A	3	38592083	NM_198056: exon28: c.5777_5780dup (p.F1928Pfs*17)	Het	Atrial fibrillation, familial, 10, [MIM:614022]; Sick sinus syndrome 1, [MIM:608567]; Ventricular fibrillation, familial, 1, [MIM:603829]; Long QT syndrome 3, [MIM:603830]; Heart block, nonprogressive, [MIM:113900]; Cardiomyopathy, dilated, 1E, [MIM:601154]; Brugada syndrome 1, [MIM:601144]; Heart block, progressive, type IA, [MIM:113900]	AD/ AR	-	NA	Likely pathogenic
20	TBX5	12	114839706	NM_000192: exon3: c.166delG	Het	Holt-Oram syndrome, [MIM:142900]	AD	-	De novo	Pathogenic

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