Table S1. A list of 14 genes with mutations identified in patients with diaphragm defect

Gene Symbol	Entrez Gene ID	Nucleotide, Amino acid	PMID	Phenotype	Human Phenotype Citations
WT1	7490	c.1097G > A,p.A366H	16932893; 18203154	Denys-Drash syndrome, CDH	Cho HY et al.Hydrothorax in a patient with Denys-Drash syndrome associated with a diaphragmatic defect. Pediatr Nephrol. 2006 Dec;21(12):1909-12.; Antonius T et al. (2008) Denys-Drash syndrome and congenital diaphragmatic hernia: another case with the 1097G > A(Arg366His) mutation. Am J Med Genet A. 2008 Feb 15;146A(4):496-9.
ZFPM2	23414	p.D98N; c. 234C>T, p.R112*	21525063; 16103912	isolated CDH; CDH with respiratory failure	Wat MJ et al. Genomic alterations that contribute to the development of isolated and non-isolated congenital diaphragmatic hernia. J Med Genet. 2011 May;48(5):299-307.; Ackerman KG et al. Fog2 Is Required for Normal Diaphragm and Lung Development in Mice and Humans. PLoS Genet. 2005 Jul;1(1):58-65.
STRA6	64220	p.G50AfsX22;P90L; P293L;T321P;T644M ;R655C	17273977	CDH, anophthalmia, congenital heart defects, alveolar capillary dysplasia, lung hypoplasia, mental retardation	Pasutto F et al. Mutations in STRA6 cause a broad spectrum of malformations including anophthalmia, congenital heart defects, diaphragmatic hernia, alveolar capillary dysplasia, lung hypoplasia, and mental retardation. Am J Hum Genet.2007 Mar;80(3):550-60.

EFNB1	1947	c. 432delG, p. L145WfsX14; c. 151_153delGTG, p. V51del; c.c.712delG, p.V238TrpfsX21	16639408; 20734337	CDH,craniofrontonasal syndrome	Vasudevan PC et al. Expanding the phenotype of craniofrontonasal syndrome: two unrelated boys with EFNB1 mutations and congenital diaphragmatic hernia. Eur J Hum Genet. 2006 Jul;14(7):884-7.; Hoque J et al. A novel EFNB1 mutation (c.712delG) in a family with craniofrontonasal syndrome and diaphragmatic hernia. Am J Med Genet A. 2010 Oct;152A(10):2574-7.
HLX	3142	c.35C>T,p.S12F; c.53C>T,p.S18L; c.517G>T,p.D173Y; c.704C>T,p.A235V	19459883	Isolated CDH	Slavotinek AM et al. Sequence variants in the HLX gene at chromosome 1q41-1q42 in patients with diaphragmatic hernia
DISP1	84976	c.4412C>G, p.A1471G	20799323	ventricular septal defect,abnormal aorta), left-sided Bochdalek CDH, and a left-sided cleft lip with bilateral cleft palate	Kantarci S et al. Characterization of the chromosome 1q41q42.12 region, and the candidate gene DISP1, in patients with CDH. Am J Med Genet A. 2010 Oct;152A(10):2493-504.
NIPBL	25836	c.5524C>T, p.R1841*	20156239	Brachmann-de Lange syndr	Hosokawa S et al. Brachmann-de Lange syndrome with congenital diaphragmatic hernia and NIPBL gene mutation. Congenit Anom (Kyoto). 2010 Jun;50(2):129-32.
PDGFRA	5156	c.C2889G,p.L967V	17568391	non-isolated CDH	Bleyl SB et al. Candidate genes for congenital diaphragmatic hernia from animal models: sequencing of FOG2 and PDGFRalpha reveals rare variants in diaphragmatic hernia patients.Eur J Hum Genet. 2007 Sep;15(9):950-8.

CHD2	1106	c.C5128T,p.R1710W	16736036	CDH	Slavotinek AM et al.Array comparative genomic hybridization in patients with congenital diaphragmatic hernia: mapping of four CDH-critical regions and sequencing of candidate genes at 15q26.1-15q26.2. Eur J Hum Genet. 2006 Sep;14(9):999-1008.
NR2F2	7026	c.G733T,p.L245F	16736036	CDH/15q	Slavotinek AM et al.Array comparative genomic hybridization in patients with congenital diaphragmatic hernia: mapping of four CDH-critical regions and sequencing of candidate genes at 15q26.1-15q26.2. Eur J Hum Genet. 2006 Sep;14(9):999-1008.
ARRDC4	91947	c.C1171T,p.R391W	16736036	CDH	Slavotinek AM et al.Array comparative genomic hybridization in patients with congenital diaphragmatic hernia: mapping of four CDH-critical regions and sequencing of candidate genes at 15q26.1-15q26.2. Eur J Hum Genet. 2006 Sep;14(9):999-1008.
LRP2	4036	c.8516_8519delTTTA,p.'	17632512	Donnai-Barrow syndrome,	Kantarci S et al. Mutations in LRP2, which encodes the multiligand receptor megalin, cause Donnai-Barrow and facio-oculo-acoustico-renal syndromes. Nat Genet. 2007 Aug;39(8):957-9.
FBN1	2200	c.T3277C, p.F1093L	12413333	Marfan Syndrome; MFS	Jacobs AM et al. A Recurring FBN1 Gene Mutation in Neonatal Marfan Syndrome. Arch Pediatr Adolesc Med. 2002 Nov;156(11):1081-5.
EFEMP2	30008	c.169G-->A, p.E57K	16685658	Cutis laxa, autosomal recess	Hucthagowder V, et al (2006) Fibulin-4: A Novel Gene for an Autosomal Recessive Cutis Laxa Syndrome. Am J Hum Genet 78(6): 1075–1080.