Supplemental Table S1. Molecular Details and Clinical Features of all five index families analysed with ES.

		Family 1 (CHT3)	Family 2 (PUV146)	Family 3 (CHT17)	Family 4 (HRZ2)	Family 5 (HRZ10)
Molecular Details	**Candidate Gene**	PKD1L1	PKD1L1	DNAH5	No Results	No Results
	Zygosity	Compound Heterozygous	Compound Heterozygous	Compound Heterozygous		
	Inheritance	Autosomal Recessive	Autosomal Recessive	Autosomal Recessive		
	gDNA location	Chr7:47944902C>T Chr7:47913548A>T	Chr7:47869647C>A Chr7:47968998delT	Chr5:13850661G>A Chr5:13882811T>A		
	Variant location	c.1543G>A, p.Gly515Arg c.3845T>A, p.Val1282Glu	c.6549G>T, p.Gln2183His c.863delA, p.Asn288Thrfs*3	c.5105C>T, p.Ser1702Phe c.3179A>T, p.Lys1060Met		
Clinical Features	**Sex**	Female	Male	Male	Female	Female
	Age of Onset	Congenital	Congenital	Congenital	Congenital	Congenital
	Primary Phenotype	Chylothorax, left Hydrops fetalis	Hydrothorax, bilateral Hydrops fetalis	Chylothorax, left	Pleural effusion, left	Chylothorax, bilateral
	Secondary Phenotype	Persistent pulmonary hypertension, respiratory failure	Severe pulmonary hypoplasia, persistent pulmonary hypertension, cardio-respiratory failure	Pneumothorax, bilateral Lymphoedema	No	Respiratory distress syndrome, cardio-respiratory failure, persistent foramen ovale
	Prenatal intervention	3x shunt insertion, thoracocentesis	3x shunt insertion	No	1x shunt insertion	No