|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Pt ID | Cardiac anomaly | Additional anomalies | Pathogenic variant(s) | Zygosity | Allele | Syndrome | Gest. at testing (wks) | Outcome |
| CHD008 | Right atrial isomerism |  | CCDC103 c.461A>C p.(His154Pro) | Hom | B | Primary ciliary dyskinesia | 19+3 | TOP |
| CHD017 | AVSD |  | ANKRD11 c.5957\_5958del p.(Arg1986Ilefs\*45) | Het | M | KBG | 20+6 | TOP |
| CHD021 | TGA, right aortic arch |  | SOS1 c.796\_797insAAG p.(Thr266delinsLysAla) | Het | M | Noonan | 28 | TOP |
| CHD035 | VSD | GI, skeletal | EVC2 c.3637\_3638insTT p.(Trp1213Phefs\*11) | Hom | B | Ellis-Van-Creveld | 20+4 | LB |
| CHD043 | Truncus arteriosus | Ventriculomegaly, DWM, CLP | CHD7 c.988C>T p.(Gln330\*) | Het | M | CHARGE | 21+3 | LB |
| CHD046 | VSD, TAPVD | CLP, collapsed stomach | CHD7 c.757del p.(Val253Cysfs\*52) | Het | M | CHARGE | 22+3 | LB |
| CHD093 | AVSD, LSVC |  | GATA4 c.220delG p.(AlaProfs\*174) | Het | M |  | 24+6 | LB |
| CHD099 | Right atrial isomerism | Ventriculomegaly | DNAH11 c.13075C>T p.(Arg4359\*) | Hom | B | Primary ciliary dyskinesia | 24+4 | TOP |
| CHD115 | TR, ECF | Multiple systemic | TCTN2 c.1506-2A>G | Hom | B | Meckel Gruber | 14+2 | SB |
| CHD112\* | HLHS | Single umbilical artery | KMT2D c.11848C>T p.(Gln3950\*) | Het | M | Kabuki | 20+6 | TOP |
| CHD118\* | TR | Increased NT, pleural effusion | RIT1 c.268A>G p.(Met90Val) dn (had subsequent NIPD) | Het | M | Noonan | 16+1 | TOP |

Table S3 – Cardiac anomalies with pathogenic variants detected on exome sequencing. [Abbreviations: AVSD= Atrioventricular septal defect; B = Biallelic CLP = Cleft lip and palate; DWM = Dandy Walker Malformation; ECF = Echogenic cardiac focus; GI = Gastrointestinal; Het = Heterozygous; Hom = Homozygous; HLHS = Hypoplastic left heart syndrome; LB = Livebirth; LSVC = Left superior vena cava; M = Monoallelic; NT = Nuchal translucency; SB=Stillbirth; TAPVD = Total anomalous pulmonary venous drainage; TGA = Transposition of the great arteries; TOP = Termination of pregnancy; TR = Tricuspid regurgitation; VSD = Ventricular septal defect] \*Variants not previously reported in the PAGE study.13