

Supplementary Table 1 Summary of previously reported studies on the 1q21.1 repeat region

Patient	Sex	Clinical manifestations	Duplication region	Duplication	Gene	Ref.
1	F	Developmental delay, craniofacial dysmorphism, congenital heart disease	chr1:148511359-148954460	335kb	<i>NBPF15, NBPF16</i>	Lijuan Zhu, 2021
2	F	Chronic depression and anxiety, facial dysmorphism	chr1:145243316-147814694	2.6Mb	<i>PRKAB2, FMO5, CHD1L, BCL9, ACP6, GJA5, GJA8, GPR89, BNBPF11, NBPF24, ITGA1, CD160</i>	Judith M.A., 2015
3	F	Congenital heart defect, mental impairment and developmental delay.	chr1:144972830-146608260	1.6 Mb	<i>HFE2HYDIN2, RBM8A, CD160, PDZK1, PIAS3, GPR89C</i>	Guowen Sun, 2014
4	F	Inattentive symptoms	chr1:146533376-147883376	1.481 Mb	<i>FMO5, CHD1L, NBPF11, PRKAB2, GJA5, GJA8</i>	Aravindhana, 2018

5	M	Facial dysmorphism	chr1:1460 23922- 147820342	1.796Mb	<i>GJA5,</i> <i>PRKAB2,</i> <i>GJA8,</i> <i>NBPF12</i>	<i>GJA8,</i> <i>CHD1L,</i> <i>NBPF11,</i>	Hongguo Zhang, 2021
6	M	Absent nasal bone, VSD, circular of umbilical cord	chr1:1466 02934- 147844778	1.242 Mb	<i>GJA5,</i> <i>PRKAB2,</i>	<i>GJA8,</i> <i>CHD1L</i>	Zhongguo Zhang, 2021
7	M	Facial dysmorphism	chr1:1450 56290- 146297463	1,241kb	<i>EFHC1,</i> <i>ATF6,</i> <i>PDZK1</i>	<i>NOS1,</i> <i>GPR89A,</i>	Harvard <i>et</i> <i>al.</i> , 2011
8	F	Refractive error, developmental delay, facial dysmorphism	chr1:1463 47156- 148575046	2227kb	<i>PRKAB2,</i> <i>CHD1L,</i> <i>ACP6,</i>	<i>FMO5,</i> <i>BCL9,</i> <i>GJA5,</i> <i>GJA8</i>	A.C Ceylan, 2019
9	F	Microcephaly and mild dysmorphic features, intellectual disability, psychomotor and language delay	chr1:145.6 32.334- 146.564.80 2	932 kb	<i>GPR89A,</i> <i>CD160,</i> <i>POLR3C</i>	<i>PDZK1,</i> <i>RNF115,</i>	Martina Busè, 2017

10	M	Dysmorphic features and psychomotor delay	chr1:145.2 91.711- 145.747.26 9	456 kb	<i>NBPF20</i> , <i>GPR89A</i> , <i>PDZK1</i> , <i>CD160</i> , <i>RNF115</i>	Martina Busè, 2017
11	F	Ventricular septal defect, developmental delay, epilepsy	chr1:1458 28373- 147924436	2.096Mb	<i>GJA5</i> , <i>FMO5</i> , <i>CHD1L</i> , <i>PRKAB2</i> , <i>BCL9</i> , <i>NBPF11</i> , <i>NBPF12</i> , <i>NBPF24</i> , <i>GJA8</i> , <i>GPR89B</i> , <i>GPR89C</i>	This study
