Supplemental Table 1. Reported uniparental isodisomy causing autosomal recessive diseases

UPiD Pattern	Parental origin	ND	Chr.	Gene	Disease	Method	Reference
s-MiRe	M		2	*TPO*	Congenital hypothyroidism	MS	Bakker et al. 2001
s-MiRe	M		4	*MTP*	Abetalipoproteinemia	MS	Yang et al. 1999
s-MiRe	M		7	*ATP6V0A4*	Distal renal tubular acidosis	SA	This study
s-MiRe	M		13	*PCCA*	Propionic acidemia	MS	Pérez et al. 2012
s-MiRe	P		2	*ABCB11*	Progressive familial intrahepatic cholestasis	MS	Giovannoni et al. 2012
s-MiRe	P		5	*MTRR*	Homocystinuria cblE type	MS	Pérez et al. 2012
s-MiRe	P		11	*HBB*	Sickle cell disease	SA	Swensen et al. 2010
s-UPiD/hD	M	M1	1	*LAMB3*	Herlitz type junctional epidermolysis bullosa	MS	Pulkkinen et al. 1997
s-UPiD/hD	M	M1	1	*LMNA*	Mandibuloacral dysplasia	MS	Bai et al. 2014
s-UPiD/hD	M	M1	2	*ALS2*	Infantile-onset ascending spastic paralysis	MS	Herzfeld et al. 2009
s-UPiD/hD	M	M1	2	*DGUOK*	Hepatocerebral mitochondrial DNA depletion syndrome	MS+SA	Haudry et al. 2012
s-UPiD/hD	M	M1	4	*FGB*	Congenital hypodysfibrinogenaemia	MS+SA	Ding et al. 2012
s-UPiD/hD	M	M1	15	*HEXA*	Tay–Sachs disease	SA	Zeesman et al. 2015
s-UPiD/hD	M	M1	15	*RECQL3*	Bloom syndrome	MS	Woodage et al. 1994
s-UPiD/hD	M	M1	16	*APRT*	Adenine phosphoribosyltransferase deficiency	MS	Ceballos-Picot et al. 2011
s-UPiD/hD	M	M1	16	*FA2H*	Spastic paraplegia type 35	SA	Soehn et al. 2016
s-UPiD/hD	M	M1	16	*FANCA*	Fanconi anemia	SA	Donovan et al. 2016
s-UPiD/hD	M	M1	16	*GALNS*	Morquio A syndrome	MS	Catarzi et al. 2012
s-UPiD/hD	M	M1	16	*MLYCD*	Malonyl CoA decarboxylase deficiency	MS	Malvagia et al. 2007
s-UPiD/hD	M	M1	17	*CTNS*	Infantile cystinosis	MS	Lebre et al. 2009
s-UPiD/hD	M	M1	22	*CYB5R3*	Recessive congenital methemoglobinemia	MS	Huang et al. 2012
s-UPiD/hD	M	M1	22	*PLA2G6*	Infantile neuroaxonal dystrophy 1	SA	Solomons et al. 2014
s-UPiD/hD	M	M2	1	*GJC2*	Pelizaeus–Merzbacher-like disease	SA	Shimojima et al. 2013
s-UPiD/hD	M	M2	2	*SFTPB*	Lethal surfactant deficiency	MS	Hamvas et al. 2009
s-UPiD/hD	M	M2	4	*MTTP*	Abetalipoproteinemia	MS	Aminoff et al. 2012
s-UPiD/hD	M	M2	4	*SGCB*	Limb-girdle muscular dystrophy 2E	SA	Cottrell et al. 2012
s-UPiD/hD	M	M2	6	*CUL7*	3M syndrome	SA	Sasaki et al. 2011
s-UPiD/hD	M	M2	9	*CHH*	Cartilage hair hypoplasia	MS	Sulisalo et al. 1997
s-UPiD/hD	M	M2	9	*FOXE1*	Syndromic congenital hypothyroidism	MS	Castanet et al. 2010
s-UPiD/hD	M	M2	16	*FANCC*	Fanconi anemia	SA	Donovan et al. 2016
s-UPiD/hD	P	M1	1	*ABCA4*	Stargardt disease	MS	Riveiro-Alvarez et al. 2007
s-UPiD/hD	P	M1	1	*P131R*	Fumarase deficiency	MS	Zeng et al. 2006
s-UPiD/hD	P	M1	1	*USH2A*	Usher syndrome type II	MS	Rivolta et al. 2002
s-UPiD/hD	P	M2	1	*CTSK*	Pycnodysostosis	MS	Gelb et al. 1998
w-UPiD	M		1	*CD45*	Severe combined immunodeficiency	SA	Roberts et al. 2012
w-UPiD	M		1	*LAMB3*	Herlitz junctional epidermolysis bullosa	MS	Castori et al. 2008
w-UPiD	M		1	*LYST*	Chediak-Higashi syndrome	MS	Dufourcq-Lagelouse et al. 1999
w-UPiD	M		1	*PEX*	Zellweger syndrome	MS	Turner et al. 2007
w-UPiD	M		3	*COL7A1*	Recessive dystrophic epidermolysis bullosa	MS	Fassihi et al. 2006
w-UPiD	M		6	*TULP1*	Cone dystrophy	MS	Roosing et al. 2013
w-UPiD	M		6	*CYP21A2*	Congenital adrenal hyperplasia	MS	Spiro et al. 1999
w-UPiD	M		7	*CFTR*	Cystic fibrosis	MS	Spence et al. 1988
w-UPiD	M		7	*CLCN1*	Recessive congenital myotonia	MS	Bulli et al. 2009
w-UPiD	M		8	*TMEM67*	Meckel–Gruber syndrome	SA	Bruechle et al. 2017
w-UPiD	M		9	*CHH*	Cartilage hair hypoplasia	MS	Sulisalo et al. 1997
w-UPiD	M		10	*HPS1*	Hermansky-Pudiak syndrome type 1	SA	Li et al. 2016
w-UPiD	M		12	*VDR*	Hereditary 1,25-dihydroxyvitamin D-resistant rickets	SA	Tamura et al. 2015
w-UPiD	M		12	*VWF*	Von Willebrand disease type 3	MS	Boisseau et al. 2011
w-UPiD	M		X	*DMD*	Duchenne muscular dystrophy	MS	Quan et al. 1997
w-UPiD	P		1	*ALPL*	Perinatal hypophosphatasia	SA	Watanabe et al. 2013
w-UPiD	P		1	*CFH*	Complement factor H deficiency	MS	Schejbel et al. 2011
w-UPiD	P		1	*GBA, MPZ*	Concurrently Gaucher disease Type 3 and Charcot-Marie-Tooth disease type I	MS	Benko et al. 2008
w-UPiD	P		1	*GNPAT*	Rhizomelic chrondrodysplasia punctata Type 2	MS	Nimmo et al. 2010
w-UPiD	P		1	*HMGCL*	3-hydroxy-3-methylglutaryl-CoA lyase deficiency	SA	Aoyama et al. 2015
w-UPiD	P		1	*LAMB3*	Herlitz junctional epidermolysis bullosa	MS	Fassihi et al. 2005
w-UPiD	P		1	*PPT1*	Infantile neuronal ceroid lipofuscinosis	SA	Niida et al. 2016
w-UPiD	P		1	*PPT1*	Neuronal ceroid lipofuscinosis type 1	SA	Travaglini et al. 2017
w-UPiD	P		1	*PRG4*	CACP syndrome	MS	Ciullini Mannurita et al. 2014
w-UPiD	P		1	*RPE65*	Retinal dystrophy	MS	Thompson et al. 2002
w-UPiD	P		1	*TRKA*	Congenital insensitivity to pain with anhidrosis	MS	Miura et al. 2000
w-UPiD	P		2	*ABCA12*	Harlequin ichthyosis	MS	Castiglia et al. 2009
w-UPiD	P		2	*CYP1B1*	Primary congenital glaucoma	MS	López-Garrido et al. 2009

w-UPiD	P	2	*DGUOK*	Mitochondrial DNA depletion syndrome	SA	Douglas et al. 2011
w-UPiD	P	2	*GGCX*	Inherited deficiency of all vitamin K-dependent coagulant factors	NGS	Dasi et al. 2016
w-UPiD	P	2	*HADHA*	Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency	MS	Baskin et al. 2010
w-UPiD	P	2	*LRP2*	Donnai–Barrow syndrome	MS	Kantarci et al. 2008
w-UPiD	P	2	*MERTK*	Retinal dystrophy	MS	Thompson et al. 2002
w-UPiD	P	2	*NEB*	Nemalin myopathy	SA	This study
w-UPiD	P	2	*UGT1A1*	Crigler–Najjar syndrome type I	MS	Petit et al. 2005
w-UPiD	P	3	*LAMB2*	Pierson syndrome	MS	Matejas et al. 2011
w-UPiD	P	5	*SMN1*	Spinal muscular atrophy	MS	Brzustowicz et al. 1994
w-UPiD	P	6	*IFNGR1*	Mendelian susceptibility to mycobacterial disease	SA	Prando et al. 2010
w-UPiD	P	6	*MUT*	Methylmalonic acidemia and agenesis of pancreatic beta cells causing diabetes mellitus	MS	Abramowicz et al. 1994
w-UPiD	P	7	*CFTR*	Cystic fibrosis	MS	Le Caignec et al. 2007
w-UPiD	P	7	*DNAH11*	Primary ciliary dyskinesia	MS	Bartoloni et al. 2002
w-UPiD	P	8	*ASAH1*	Spinal muscular atrophy and progressive myoclonic epilepsy	MS	Giráldez et al. 2015
w-UPiD	P	8	*CYP11B1*	Congenital adrenal hyperplasia	SA	Matsubara et al. 2014
w-UPiD	P	8	*LPL*	Lipoprotein lipase deficiency	MS	Benlian et al. 1996
w-UPiD	P	12	*SUOX*	Isolated sulfite oxidase deficiency	SA	Cho et al. 2013
w-UPiD	P	13	*SACS*	Autosomal recessive spastic ataxia of Charlevoix Saguenay	MS	Anesi et al. 2011
w-UPiD	P	16	*ABCA3*	Lethal surfactant deficiency	MS	Hamvas et al. 2009
w-UPiD	P	16	*FA2H*	Spastic paraplegia type 35	SA	Soehn et al. 2016
w-UPiD	P	16	*FANCA*	Fanconi anemia	SA	Donovan et al. 2016
w-UPiD	P	17	*ITGB4*	Junctional epidermolysis bullosa with pyloric atresia	MS	Natsuga et al. 2010
w-UPiD	P	22	*ARSA*	Metachromatic leukodystrophy	SA	Niida et al. 2012

Chr., chromosome; M, maternal; M1, meiosis I; M2, meiosis II; MS, Microsatellite analysis; ND, nondisjunction; NGS, next generation sequencing; P, paternal; SA, SNP Array; s-MiRe, segmental UPiD by mitotic recombination; s-UPiD/hD, segmental UPiD with uniparental heterodisomy; UPiD, uniparental isodisomy; w-UPiD, whole chromosomal UPiD.

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Supplemental Table 2. Distribution of the informative SNP on chromosome 7 of the patient 2

Cytogenetic bands	Chromosomal Position (hg19)	Informative SNP for heterozygous from both parents; F(AA)/M(BB)/Pt(AB)		Informative SNP for maternal UPiD; F(AA)/M(AB)/Pt(BB)	
p22.3	5,0814 ~ 2,800,000	33		1	
p22.2	2,800,000 ~ 4,500,000	31		0	
p22.1	4,500,000 ~ 7,300,000	16		0	
p21.3	7,300,000 ~ 13,800,000	143		0	
p21.2	13,800,000 ~ 16,500,000	70		0	
p21.1	16,500,000 ~ 20,900,000	96		0	
p15.3	20,900,000 ~ 25,500,000	101		0	
p15.2	25,500,000 ~ 28,000,000	13		1	
p15.1	28,000,000 ~ 28,800,000	28		0	
p14.3	28,800,000 ~ 35,000,000	106	1007 (98.2%)	0	3 (0.2%)
p14.2	35,000,000 ~ 37,200,000	75		0	
p14.1	37,200,000 ~ 43,300,000	80		0	
p13	43,300,000 ~ 45,400,000	2		0	
p12.3	45,400,000 ~ 49,000,000	75		1	
p12.2	49,000,000 ~ 50,500,000	19		0	
p12.1	50,500,000 ~ 54,000,000	75		0	
p11.2	54,000,000 ~ 58,000,000	24		0	
p11.1	58,000,000 ~ centromere	No probe area			
q11.1	centromere ~ 61,700,000				
q11.21	61,700,000 ~ 67,000,000	22		0	
q11.22	67,000,000 ~ 72,000,000	0		134	
q11.23	72,000,000 ~ 77,500,000	0		22	
q21.11	77,500,000 ~ 86,400,000	1		268	
q21.12	86,400,000 ~ 88,200,000	1		37	
q21.13	88,200,000 ~ 91,000,000	0		34	
q21.2	91,000,000 ~ 97,800,000	2		141	
q21.3	97,800,000 ~ 98,000,000	0		1	
q22.1	98,000,000 ~ 103,800,000	1		59	
q22.2	103,800,000 ~ 104,500,000	0		17	
q22.3	104,500,000 ~ 107,400,000	0		60	
q31.1	107,400,000 ~ 114,600,000	4		124	
q31.2	114,600,000 ~ 117,400,000	0	18 (1.8%)	34	1860 (99.8%)
q31.31	117,400,000 ~ 121,000,000	1		27	
q31.32	121,000,000 ~ 123,800,000	0		13	
q31.33	123,800,000 ~ 127,000,000	1		87	
q32.1	127,000,000 ~ 129,200,000	0		10	
q32.2	129,200,000 ~ 130,400,000	1		19	
q32.3	130,400,000 ~ 132,600,000	1		111	
q33	132,600,000 ~ 138,200,000	3		164	
q34	138,200,000 ~ 143,100,000	1		81	
q35	143,100,000 ~ 147,900,000	1		93	
q36.1	147,900,000 ~ 152,600,000	0		118	
q36.2	152,600,000 ~ 155,000,000	0		94	
q36.3	155,000,000 ~ 159,130,000	0		92	
Total SNP number		1025		1863	

F, father; M, mother; Pt, patient; UPiD, uniparental isodisomy

NOTE: The informative SNP patterns clearly changes between q11.21 and q11.22 as shown in Fig. 2B. It was impossible to determine whether the UPiD within a distal region of short arm in the figure is real or not from the informative SNP patterns.

Supplemental Table 3. Distribution of the informative SNP on chromosome 15 of the patient 3

Cytogenetic bands	Chromosomal Position (hg19)	Informative SNP for maternal UPiD; F(AA)/M(AB)/Pt(BB)	Maternal UPiD SNP density (SNP/Mb)	Informative SNP for maternal UPiD or UPhD; F(AA)/M(BB)/Pt(BB)	Maternal UPiD or UPhD SNP density (SNP/Mb)	distance (Mb)
q11	20,700,000 ~ 25,700,000	4	0.8	18	3.6	5.0
q12	25,700,000 ~ 28,000,000	5	2.2	31	13.5	2.3
q13.2	28,000,000 ~ 31,200,000	5	1.6	10	3.1	3.2
q13.3	31,200,000 ~ 33,600,000	14	5.8	9	3.8	2.4
q14	33,600,000 ~ 40,000,000	185	**28.9**	67	10.5	6.4
q15	40,000,000 ~ 44,800,000	11	2.3	3	0.6	4.8
q21.1	44,800,000 ~ 49,500,000	10	2.1	39	8.3	4.7
q21.2	49,500,000 ~ 52,900,000	5	1.5	38	11.2	3.4
q21.3	52,900,000 ~ 59,000,000	12	2.0	1	0.2	6.1
q22.2	59,000,000 ~ 63,700,000	10	2.1	45	9.6	4.7
q22.3	63,700,000 ~ 67,500,000	2	0.5	31	8.2	3.8
q23	67,500,000 ~ 72,700,000	3	0.6	14	2.7	5.2
q24.1	72,700,000 ~ 75,200,000	1	0.4	8	3.2	2.5
q24.3	75,200,000 ~ 78,300,000	3	1.8	16	5.2	3.1
q25.1	78,300,000 ~ 81,700,000	7	2.1	22	6.5	3.4
q25.2	81,700,000 ~ 85,200,000	2	0.6	10	2.9	3.5
q25.3	85,200,000 ~ 89,000,000	122	**32.1**	42	11.1	3.8
q26.1	89,000,000 ~ 94,300,000	229	**43.2**	75	14.2	5.3
q26.2	94,300,000 ~ 98,500,000	164	**39.0**	68	16.2	4.2
q26.3	98,500,000 ~ 102,500,000	88	**22.0**	52	13.0	4.0
Total SNP number		882		1556		

F, father; M, mother; Pt, patient; UPhD, uniparental heterodisomy; UPiD, uniparental isodisomy

NOTE: Consistent with the Fig. 2C, the density of UPiD is high at q14 and q25.3 to qter. It was impossible to determine whether the UPiD at q22 and q24.1 in the figure is real or not from the informative SNP patterns.