Table S1. Summary of reported cases of maternal uniparental disomy of chromosome 6 in the literature.

First Author, Year	Sex	UPD Type	Monogenic Mutation	Variant Consequences in a Patient	IUGR (HP:0001511)	Other Prenatal Findings	Gestational Age at birth (whole completed weeks)	Birth Weight (g)	Weight z-score	SGA, AGA OR LGA	Failure to Thrive (FTT)	Postnatal Phenotype other than correlating to the mutation
Our case	F	Isodisomy	DIAPH2 c.881T>A p.(Leu294His), heterozygous	NR	Yes	Polyhydramnios HP:0001561;	35	1450	-2,7	SGA	Yes	Sacrococcygeal teratoma HP:0030736; Delayed speech and language development HP:0000750; Autism Spectrum Disorder HP:0000717; Triangular face HP:0000325; Prominent forehead HP:0011220; Full lips HP:0012471; Reduced subcutaneous adipose tissue HP:0003758; Genu valgum HP:0002857; Pes planus HP:0001763;
Li, J. W., et al. (2024).	M	Hetero/Isodisomy	Not detected	NR	Yes	NR	32	1300	-1,5	SGA	Yes	Gastrostomy tube feeding in infancy HP:0011471; Allergic rhinitis HP:0003193; High palate HP:0000218;
Li, J. W., et al. (2024).	F	Isodisomy	SCUBE3 c.2330T > C (p. Phe-777Ser), homozygous	Short stature HP:0004322;	Yes	NR	36	1200	-3,9	SGA	Yes	none
Jiang, Y., et al. (2024).	M	Hetero/Isodisomy	Not detected	NR	Yes	NR	termination of pregnancy	no data	no data	no data	NR	NR
Jiang, Y., et al. (2024).	F	Hetero/Isodisomy	Not detected	NR	Yes	Fetal abdominal cyst HP:4000139	termination of pregnancy	no data	no data	no data	NR	NR
Zhang, P., et al. (2021).	M	Isodisomy	IFNGR1 c.861+2T>A, homozygous	AR complete IFN-γR1 deficiency	Yes	NR	40	2450	-2,8	SGA	No	none
Souzeau, E., et al. (2018).	F	Isodisomy	TULP1 c.524dupC, p.(Pro176ThrfsTer7), homozygous; RP1 (c.1267_1656delinsA, p.(Ala423AsnfsTer11)), heterozygous	Rod-cone dystrophy HP:0000510	Yes	Oligohydramnios HP:0001562;	37	1640	-3,3	SGA	No	none
Kerr, E. R., et al. (2018).	F	Isodisomy	Not detected	NR	No	Nuchal cord HP:0012498	32	1304	-1,3	SGA	No	Intraventricular hemorrhage HP:0030746; Preauricular pit HP:0004467;
Leung, W. C., et al. (2017).	F	Hetero/Isodisomy	mosaic placenta 47,XX, +6[12]/46,XX[19]	Confined placental mosaicism	Yes	Oligohydramnios HP:0001562;	34	1115	-3	SGA	No	none
Leung, W. C., et al. (2017).	F	Heterodisomy	mosaic placenta 47,XX, +6[14]/46,XX[16]	Confined placental mosaicism	Yes	NR	32	890	-2,6	SGA	No	none
Eggermann, T., et al. (2017).	F	Hetero/Isodisomy	46,XX (FISH: 98/100 normal, 2x monosomy 6)	NR	Yes	Bradycardia HP:0001662	27	650	-1,4	SGA	Yes	Clinodactyly of the 5th finger HP:0004209; Restlessness HP:0000711; Macrotia HP:0000400; Triangular face HP:0000325; Frontal bossing HP:0002007; Mandibular prognathia HP:0000303
Eggermann, T., et al. (2017).	M	Hetero/Isodisomy	CYP21A2: exon 1–8 deletion, homozygous	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency OMIM:201910;	Yes	Oligohydramnios HP:0001562;	30	1100	-1,1	AGA	Yes	Short palpebral fissure HP:0012745; Macrotia HP:0000400; Clinodactyly of the 5th finger HP:0004209; Pes valgus HP:0008081; Pes planus HP:0001763; Macrotia HP:0000400
Lazier, J., et al. (2016).	M	Isodisomy	Not detected	NR	Yes	Placental dysfunction	28	680	-1,7	SGA	Yes	Ambiguous genitalia, male HP:0000033; Persistent Müllerian duct syndrome ORPHA:2856; Camptodactyly of finger HP:0100490; Perineal hypospadias HP:000005; Inguinal hernia HP:0000023; Patent ductus arteriosus HP:0001643; Thick septum pellucidum HP:0007375;
Takada, H., et al. (2014).	F	Isodisomy	WASP c.1276_1285 del_GCCCCTG GTG: p.Ala426GlyfsX15, heterozygous	Wiskott-Aldrich syndrome OMIM: 301000;	Yes	NR	29	641	-2	SGA	No	none
Roosing, S., et al. (2013).	M	Isodisomy	TULP1 c.1258C A; p.Arg420Ser, homozygous	Cone dystrophy HP:0008020;	Yes	NR	40	no data	no data	no data	No	none
Poke, G., et al. (2013).	F	Isodisomy	Not detected	NR	Yes	Decreased fetal movement HP:0001558; Placental insufficiency ORPHA:439167; Abnormal placenta morphology HP:0100767;	35	1200	-3,5	SGA	Yes	Dysphagia HP:0002015; Gastroesophageal reflux HP:0002020; Global developmental delay HP:0001263
Begemann, M., et al., (2012).	F	Heterodisomy	Not detected	NR	Yes	Decreased fetal movement HP:0001558; Oligohydramnios HP:0001562;	34	1030	-3,2	SGA	Yes	Midface retrusion HP:0011800; Prominent forehead HP:0011220; Relative macrocephaly HP:0004482; Hypotonia HP:0001252; Motor delay HP:0001270; Umbilical hernia HP:0001537;
Sasaki, K., et al. (2011).	M	Hetero/Isodisomy	CUL7: c.2975G > C (p.R992P), homozygous	3M-syndrome OMIM: 273750;	Yes	NR	36	1000	-4,6	SGA	Yes	Mild mental retardation HP:0001256; Inguinal hernia HP:0000023; Hydrocele testis HP:0000034; Ventriculomegaly HP:0002119; Feeding difficulties in infancy HP:0008872; Hypotonia HP:0001252; Hypospadias HP:0000047; Triangular face HP:0000325; Malar flattening HP:0000272; Epicanthus HP:0000286; Short nose HP:0003196; Anteverted nares HP:0000463; Full lips HP:0012471; Long philtrum HP:0000343; Hypermobility of toe joints HP:0010510; Slender long bone HP:0003100; Pointed chin HP:0000307;
Salahshourifar, I., et al. (2010).	M	Heterodisomy	Not detected	NR	No	Maternal hypertension HP:0008071	40	3700	0,2	AGA	No	Orofacial cleft HP:0000202;
Gümüş, H., et al. (2010).	M	Isodisomy	MOCS1: c.217C>T (p.Arg73Trp), homozygous	Molybdenum cofactor deficiency, OMIM:252150;	No	Dandy-Walker malformation HP:0001305;	40	3750	0,3	AGA	No	Triangular face HP:0000325; Anteverted nares HP:0000463; Microcephaly HP:0000252; Motor delay HP:0001270;
Parker, E. A., et al. (2006).	M	Isodisomy	48,XXY, mar[30]/ 47,XXY[20], CYP21A2 whole gene deletion, homozygous, UPD X(mat)	Klinefelter syndrome; Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency OMIM:201910;	Yes	Oligohydramnios HP:0001562	36	2060	-1,8	SGA	Yes	Persistent pulmonary hypertension of the newborn HP: 0002092; Microcephaly HP:0000252; Patent ductus arteriosus HP:0001643;

Cockwell, A. E., et al. (2006).	M	Heterodisomy	mos46,XY[13]/47,XY, + 6[12]	NR	NR	Prenatal death HP:0034241	23	no data	no data	no data	NR	Prenatal death HP:0034241; Atrioventricular canal defect HP:0006695; Omphalocele HP:0001539;
Spiro, R. P., et al. (1999).	F	Isodisomy	CYP21A2: (p.I172N), homozygous	Adrenal hyperplasia, congenital, due to 21-hydroxylase deficiency OMIM:201910;	Yes	NR	37	1410	-3,9	SGA	No	Motor delay HP:0001270; Chronic otitis media HP:0000389;
van den Berg-Loonen (1996).	M	Isodisomy	Not detected	NR	Yes	NR	40	1500	-5,3	SGA	Yes	Sarcoidosis ORPHA:797; Renal insufficiency HP:0000083;

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