Supplementary Table 3. Clinical features of our two patients with UPD(16)mat and phenotypical comparison between patients with UPD(16)mat and 11p15 LOM or UPD(7)mat.

Patient	UPD(16)mat					SRS		P value[a]	
	Patient 1	Patient 2	Our patients	Previously reported patients	Total	11p15 LOM	UPD(7)mat	UPD(16)mat vs. 11p15 LOM	UPD(16)mat vs. UPD(7)mat
Sex	Male	Female							
Karyotype	46,XY	46,XX							
Present age (years)	5	11							
Gestational age (weeks)	27.3	29.3	27.3, 29.3	35.1 (28.0~40.0) (n=33)	35.0 (27.3~40.0) (n=35)	38.0 (34.4~40.0) (n=36)	38.0 (34.6~40.0) (n=9)		
Preterm birth	+	+	2/2	23/36[b]	25/38				
Birth length in cm (SDS)[c]	31.0 (–1.96)	33.0 (–2.38)	(–2.38, –1.96)	…	…	(–4.13 ± 2.01) (n=31)	(–3.18 ± 1.16) (n=9)		
Birth weight in g (SDS)[c]	698 (–2.38)	806 (–2.60)	(–2.60, –2.38)	…	…	(–3.50 ± 0.85) (n=42)	(–2.90 ± 0.64) (n=9)		
Birth OFC in cm (SDS)[c]	23.0 (–1.02)	25.2 (–0.84)	(–1.02, –0.84)	…	…	(–0.54 ± 1.22) (n=29)	(–1.44 ± 0.47) (n=9)		
Height at 24 months in cm (SDS)[d,e]	71.7 (–5.24)	72.6 (–3.98)	(–5.24, –3.98)	…	…	…	…		
BMI at 24 months (SDS)[d,e]	–2.68	–2.14	–2.68, –2.14	…	…	…	…		
Present height in cm (SDS)[d]	89.9 (–4.24)	133.3 (–1.72)	(–4.24, –1.72)	…	…	(–3.58 ± 1.65) (n=35)	(–3.77 ± 1.13) (n=9)		
Present weight in kg (SDS)[d]	11.0 (–5.03)	28.4 (–1.48)	(–5.03, –1.48)	…	…	(–3.15 ± 1.16) (n=32)	(–2.77 ± 0.76) (n=9)		
GH treatment	3 years~	3 years~							
SGA[f]	+	+	2/2	24/38	26/40	43/43	9/9	**0.000**	**0.045**
Postnatal growth failure[e,g]	+	+	2/2	…	…	29/35	8/9		
Relative macrocephaly at birth[h]	–	+	1/2	…	…	29/29	7/9		
Protruding forehead	+	+	2/2	…	…	31/37	7/9		
Body asymmetry	–	–	0/2	…	…	30/37	3/9		
Feeding difficulties and/or low BMI	+	+	2/2	…	…	16/34	6/9		
NH-CSS	4/6	5/6							
Triangular face	+	+	2/2	…	…	42/43	8/9		
Fifth finger clinodactyly	–	+	1/2	…	…	29/37	5/9		
Fifth finger brachydactyly	–	+	1/2	…	…	30/38	2/9		
Congenital heart disease	VSD	–	1/2	10/31	11/33	8/145	0/17	**0.000**	**0.009**
Development									
Motor developmental delay	+	–	1/2	…	…	18/37	6/9		
Age at head control (months)[i]	12	4							
Age at sitting without support (months)[i]	17	7							
Age at walking without support (months)[i]	21	15							
Speech delay	+	–	1/2	…	…	8/31	6/9		
IQ/DQ (age at examination)	51 (3 years)	67 (6 years)		…	…				
Hypospadias and/or cryptorchidism	+	NA	1/1	6/17	7/18	12/22	2/7	0.360	1.000
Other features	Tube feeding, Retinopathy of prematurity	–							
Paternal age at childbirth (years)	40	30	30, 40	35, 36 (n=2)	35.5 (30~40) (n=4)	32 (19~52) (n=24)	35 (27~48) (n=9)		
Maternal age at childbirth (years)	44	36	36, 44	34 (21~42) (n=31)	34 (21~44) (n=33)	32 (19~43) (n=25)	33 (25~42) (n=9)		
Paternal height in cm (SDS)	166.0 (–0.82)	170.0 (–0.14)	…	…	…	…	…		
Maternal height in cm (SDS)	138.0 (–3.83)	150.0 (–1.54)	…	…	…	…	…		
Reference				[1–3]		[4–6][j]	[4–6][j]		

UPD(16)mat: maternal uniparental disomy of chromosome 16; 11p15 LOM: loss of methylation on chromosome 11p15; UPD(7)mat: maternal uniparental disomy of chromosome 7; SRS: Silver-Russell syndrome; SDS: SD score; OFC: occipitofrontal circumference; BMI: body mass index; GH: growth hormone; SGA: small for gestational age; NH-CSS: Netchine-Harbison clinical scoring system; VSD: ventricular septal defect; DQ: developmental quotient; NA: not applicable.

[a] Because there was no individual clinical data about SRS patients in the literature,[4] we performed statistical analyses between patients with UPD(16)mat and SRS only about frequencies of SGA, congenital heart diseases and hypospadias and/or cryptorchidism. Significant p values (<0.05) are boldfaced.

[b] In addition to 33 previously reported UPD(16)mat patients with information about gestational age, three term UPD(16)mat patients were reported in the literature.

[c] Birth length, weight and OFC were evaluated by the sex- and the gestational age-matched Japanese reference data (http://jspe.umin.jp/medical/chart_dl.html).

[d] Postnatal height, BMI and weight were evaluated by the sex- and the age-matched Japanese reference data (http://jspe.umin.jp/medical/chart_dl.html).

[e] If we did not get information at 24 ±1 months, we used the data at the nearest measure available older than 25 months.

[f] Birth length and/or birth weight ≤–2 SDS. For previously reported UPD(16)mat patients with information about percentile without SDS, patients whose birth length and/or birth weight ＜3rd centile or 5th centile were classified as SGA. For previously reported UPD(16)mat patients without information about SDS nor percentile, patients whose birth length and/or birth weight were ≤–2 SDS using the sex- and the gestational age-matched Japanese reference data (http://jspe.umin.jp/medical/chart_dl.html)

were classified as SGA.

[g] Height at 24 ±1 months ≤–2 SDS or height ≤–2 SDS below mid-parental target height. Mid-parental target height was calculated as follows: ((father's height+mother's height)/2) +6.5 cm for boys and –6.5 cm for girls.

[h] Head circumference at birth ≥1.5 SDS above birth length and/or weight SDS.

[i] Corrected age.

[j] All the clinical data was referred from [4], except the frequency of congenital heart diseases (from [5]) and the frequency of hypospadias and/or cryptorchidism (from [6]).

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