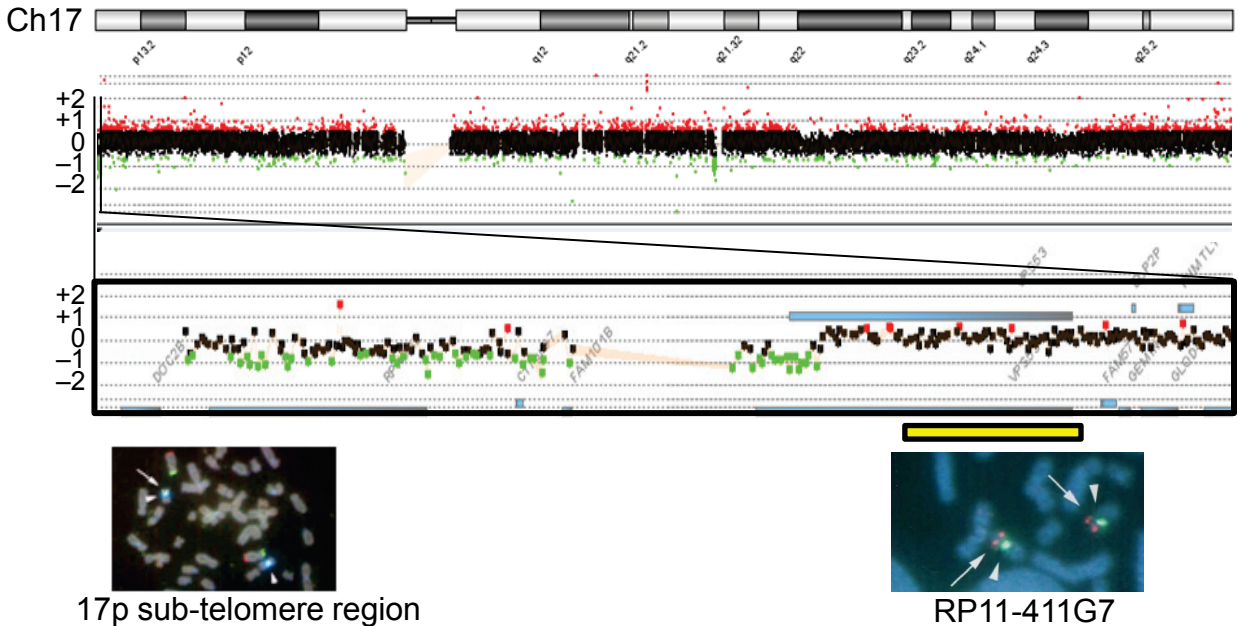


Supplemental Material to:

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Paternal uniparental disomy 14 and related disorders: Placental gene expression analyses
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Supplemental Figure 1. Array CGH and FISH analysis for the terminal 17p portion in case 4. In CGH analysis, the black, the red, and the green dots denote signals indicative of the normal, the increased ($> +0.5$), and the decreased (< -1.0) copy numbers, respectively. In FISH analysis, the 17p sub-telomeric probe detects only a single faint yellow signal (an arrow), whereas the RP11-411G7 probe identifies two red signals (arrows). The CEP17 probe for 17p11.1 is used as an internal control (arrowheads). The extra green and the red signals on the left panel are derived from the 8p probe and 8q probe, respectively.

Supplemental Table 1. Summary of clinical features

	Case 1	Case 2	Case 3	Case 4
Sex	Female	Male	Male	Female
Karyotype	46,XX,upd(14)pat	46,XY,upd(14)pat	46,XY ^c	46,XX,der(17)t(14;17)(q31;p13)pat
Gestational age (wks)	36	37	30	40
Birth length (cm) (SD) ^a	45.0 (−0.8)	48.0 (±0)	N.E.	50.5 (+0.9)
Birth weight (kg) (SD) ^a	3.4 (+2.5)	2.7 (−0.4)	2.0 ^d	3.3 (+0.6)
Upd(14)pat phenotype	Typical	Typical	Typical	Apparently absent
Polyhydramnios	+	+	+	−
Placental weight (g) (%) ^b	970 (198)	1384 (262)	642 (163)	550 (104)
Characteristic face	+	+	+	−
Bell-shaped thorax	+	+	+	−
Abdominal wall defect	+	+	+	+ (omphalocele)
Non-upd(14)pat phenotype	−	−	−	+ ^e
References	1, 2	2, 3	4	This report

^a Assessed by the gestational age- and sex-matched Japanese reference data from the Ministry of Health, Labor, and Welfare (<http://www.e-stat.go.jp/SG1/estat/eStatTopPortal.do>).

^b Assessed by the gestational age-matched Japanese reference data for placental weights.^{5,6}

^c Positive for a 108,768 bp microdeletion involving *DLK1*, the IG-DMR, the *MEG3*-DMR, and *MEG3* of maternal origin.

^d No gestational age-matched growth reference for neonates born at 30 weeks of gestational age.

^e Non-specific facial features and cardiomegaly and hepatomegaly of unknown cause.

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Supplemental Table 2. The results of microsatellite analysis

Maker	Locus	Case 1				Case 2				Case 4			
		Mother	Proband	Father	Assessment	Mother	Proband	Father	Assessment	Mother	Proband	Father	Assessment
<i>D14S609</i>	14q11.2	170	162/166	162/166	Heterodisomy	170/174	170	170	N.I.				
<i>D14S261</i>	14q11.2	189/191	189	189	N.I.	165	165	165/189	N.I.
<i>D14S80</i>	14q12	97/105	103	97/103	Isodisomy	97	97	97	N.I.
<i>D14S608</i>	14q12	212/222	208	208/212	Isodisomy	200	194	194/210	Isodisomy
<i>D14S75</i>	14q13.3	190/196	190	190	N.I.	182/196	180	180/190	Isodisomy
<i>D14S566</i>	14q21.2	122/132	144	132/144	Isodisomy	128/132	128	128/132	N.I.
<i>D14S136</i>	14q22.1	151	151	151	N.I.	151	151	151	N.I.
<i>D14S1152</i>	14q21.3	203	203	203	N.I.	203	203	203	N.I.
<i>D14S980</i>	14q22.3	153/167	167	157/167	N.I.	167/173	165	161/165	Isodisomy
<i>D14S63</i>	14q23.2	203/209	207	207	Isodisomy	203/207	203	203	N.I.
<i>D14S588</i>	14q24.2	114/122	114	110/114	N.I.	114/126	114	114/122	N.I.
<i>D14S1025</i>	14q24.3	142/144	144	144/146	N.I.	140/142	148	144/148	Isodisomy
<i>D14S617</i>	14q32.12	164	138	134/138	Isodisomy	139/169	143	143/165	Isodisomy
<i>D14S250</i>	14q32.2	162/164	160	160/164	Isodisomy	158	158	158/166	N.I.	154/158	158/162	160/162	Biparental
<i>D14S1006</i>	14q32.2	126/144	126	126/140	N.I.	126/138	126	126/138	N.I.	126/136	126	126/144	N.I.
<i>D14S985</i>	14q32.2	136	136	136	N.I.	136/138	132	132/134	Isodisomy
<i>D14S1010</i>	14q32.33	138/146	146	142/146	N.I.	134/142	142	142/144	N.I.
<i>D14S292</i>	14q32.33	111/123	123	111/123	N.I.	105/111	109	107/109	Isodisomy
<i>D14S1007</i>	14q32.33	113	123	113/123	Isodisomy	119	119	119	N.I.	111/119	119/123	111/123	Biparental

Abbreviations, N.I.: not informative

Supplemental Table 3. Primers utilized in the present study

	Forward primer	Reverse primer	AT
<Microsatellite analysis>			
<i>D14S609</i>	AGCTCTAACAGTTTTGTCATGG	TATGTGCCCTAAAGGAAGCA	57
<i>D14S261</i>	GGCAGTATGACCCAGTTACAGAT	ATTTTGAAGCCCCAGGAA	57
<i>D14S80</i>	CTTTCCTTGTTTTATAAGGCTG	CAACTTAGTGTTTCATTGATGCA	55
<i>D14S608</i>	TAAAGGTTTATCCATGCTGTAGC	ACGTGGTACAGGTAGATAAATGG	55
<i>D14S75</i>	TGTCCCCAGGTGTTGA	CCAAGTGGCCTTGCTC	57
<i>D14S566</i>	GCCACTGCACTTCTGCCTGG	CCAAGATACATTCCTGATTGAG	57
<i>D14S136</i>	TCAGAGTCGATCAAAATATGT	GATGTGAGTTTGAAAAGCAG	57
<i>D14S1152</i>	GATGTGCCAAGCACTGTTGTA	TCATACACTTCCATAGCAGCG	57
<i>D14S980</i>	CTGGGCAACAAGAGTGG	GAAGCGGGACAATTCTCTAAG	57
<i>D14S63</i>	GGCCAGGTTTCAATCAGTTT	GCCAGAGAGCCACACTGTAT	57
<i>D14S588</i>	GCCGAAAGAAAGAAAAAAGG	CGAATGCATACTTGCTGTTG	55
<i>D14S1025</i>	GATGGCACCCTGCAC	AGGAAATTAACATTTTCTTACAGC	57
<i>D14S617</i>	TTTTAGGTGGCCACCATCTA	CCAGTTTAGGCAACAGAACA	55
<i>D14S250</i>	GAAACTGGAACCACTGTGC	ACCCCTGCATTGTTTGAG	55
<i>D14S1006</i>	TTCCACAGGGCAAGCAGTA	TTCTGGCAAAACCCAACC	57
<i>D14S985</i>	CAGTGTGACCTTAAACAAGTCG	CCTGTGGGGTAGATACACGA	57
<i>D14S1010</i>	AGATTCTGGACTTGCCAAC	GTAGTAGTCAGGGCTTCCTAGAG	55
<i>D14S292</i>	CTGTGTGGTGCATCAATG	CATGAAGGCAGCCTCA	55
<i>D14S1007</i>	AGCTCCTATATGTCTTCACACAG	CTCCATTCCCATACGTCC	55
<Methylation analysis>			
CG4	TTTTATTATTGAATTGGGTTTGTTAGT	ACAATTCCTACTACAAAATTTCAACA	57
CG6	GTAAAGAGTTTGTGGATTTGTGAGAAATG	CTAAAAATCACCAAACCCATAAAATCAC	57
CG7	TTGTGTTTGAATTTATTTTGT	CCCCAAATTCATAACAAATTA	57
<Quantitative real-time PCR for <i>RTL1</i> >			
3'RACE primer for synthesizing cDNA	CTGATCTAGAGGTACCGGATCCGACTCGAGTCGACATCGTTTTTTTTTTTTTTTTTTT.		
q-PCR <i>RTL1</i>	TGCCACTTACCAGACTTGCACAGCAAAGAG	AGAGGTACCGGATCCGACTCGAGTCGACAT	70