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Corrigendum to: Clinical spectrum and management of imprinting disorders

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The authors wish to correct the legends of tables 1 and 2 as follows:

Table 1: Overview on the currently known imprinting disorders, their major molecular findings and main clinical features. (GOM, gain of methylation; IUGR, intrauterine growth restriction; MLID, multilocus imprinting distur-

bance; LOM, loss of methylation; PNGR, postnatal growth restriction; PTH, parathyroid hormone; upd, uniparental disomy)

Table 2: The recently consented clinical diagnostic criteria for SRS, BWS and iPPSDs (4) (5) (6) which might help to decide on the molecular diagnostic testing procedure. For the established diagnostic criteria for PWS and AS we refer to GeneReviews. (PTH parathyroid hormone, TSH thyroid stimulating hormone)

In addition, “parental” was incorrectly named as “paternal” in several places in table 1. A corrected version of table 1 is provided.

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Table 1: Overview on the currently known imprinting disorders, their major molecular findings and main clinical features. (GOM, gain of methylation; IUGR, intrauterine growth restriction; MLID, multilocus imprinting disturbance; LOM, loss of methylation; PNGR, postnatal growth restriction; PTH, parathyroid hormone; upd, uniparental disomy).

Imprinting disorder OMIM#	Prevalence	Chromosome	Molecular defects (frequency)	Recurrence risk	MLID	Main clinical features
Transient neonatal diabetes mellitus (TNDM) 601410	1/300,000	6q24	upd(6)pat (41 %) dup(6q24)pat (33 %) <i>PLAGL1</i> :alt-TSS-DMR, LOM (26 %)	low familial structural variants possible in case of MLID: autosomal recessive <i>ZFP57</i> mutations	50 %	IUGR, transient diabetes mellitus, hyperglycaemia without ketoacidosis, macroglossia, abdominal wall defects
Silver–Russell syndrome (SRS) 180860	1/75,000–1/100,000	7	upd(7)mat (7–10 %) upd(11p15)mat (1 case) dup(11p15)mat (<1 %) <i>H19/IGF2</i> :IG-DMR, LOM (40 %) <i>CDKN1C</i> , <i>IGF2</i> , <i>HMGGA2</i> , <i>PLAG1</i> point mutations	only one familial translocation reported not increased familial structural variants possible in case of MLID: maternal effect variants possible (familial) CNVs, SNVs in cis causing LOM reported in rare cases autosomal-dominant inheritance with parental imprinting	1 case 10 %	IUGR, PNGR, relative macrocephaly at birth, body asymmetry, prominent forehead, feeding difficulties
Birk–Barel syndrome 612292	unknown	8q24.3	<i>KCNK9</i> point mutations	autosomal-dominant inheritance with parental imprinting		Intellectual disability, hypotonia, dysmorphism
Beckwith–Wiedemann syndrome (BWS) 130650	1/15,000	11p15.5	upd(11p15)pat (20 %) dup(11p15)pat (2–4 %) <i>H19/IGF2</i> :IG-DMR, GOM (4 %) <i>KCNQ1OT1</i> :TSS-DMR, LOM (50 %) <i>CDKN1C</i> point mutations (5 %) sporadic; familial 25–50 %	no familial structural variants possible OCT4 binding site SNVs in 10 %, might be heritable in case of MLID: maternal effect variant autosomal-dominant inheritance with parental imprinting	25 %	Macroglossia, exomphalos, lateralized overgrowth, Wilms tumor or nephroblastomatosis, hyperinsulinism, adrenal cortex cytomegaly, placental mesenchymal dysplasia, pancreatic adenomatosis
Temple syndrome (TS14) 616222	unknown	14q32	upd(14)mat (70 %) del(14q32)pat (10 %) <i>MEG3/DLK1</i> :IG-DMR, LOM (20 %)	familial structural variants possible familial structural variants possible low	single cases	IUGR, PNGR, neonatal hypotonia, feeding difficulties in infancy, truncal obesity, scoliosis, precocious puberty, small feet and hands
Kagami–Ogata syndrome (KOS14) 608149	unknown	14q32	upd(14)pat (70 %) del(14q32)mat (20 %) <i>MEG3/DLK1</i> :IG-DMR, GOM (10 %)	familial structural variants possible familial structural variants possible low		IUGR, polyhydramnion, abdominal wall defects, bell-shaped thorax, coat-hanger ribs

Table 1 (continued)

Imprinting disorder OMIM#	Prevalence	Chromosome	Molecular defects (frequency)	Recurrence risk	MLID	Main clinical features
(familial) Central Precocious Puberty (CPPB)	unknown	14q32	<i>DLK1</i> point mutations	autosomal-dominant inheritance with parental imprinting		Central precocious puberty
Prader-Willi syndrome (PWS) 176270	1/25,000–1/10,000	15q11q13	del(15q11q13)pat (70–75 %) upd(15)mat (25–30 %) <i>SNURF</i> :TSS-DMR, GOM (1 %)	familial structural variants possible familial structural variants possible 0–15 % due to an IC deletion	1 case	PNGR, Intellectual disability, neonatal hypotonia, hypogenitalism, hypopigmentation, obesity, hyperphagia
Angelman syndrome (AS) 105830	1/20,000–1/12,000	15q11q13	del(15q11q13)mat (75 %) upd(15)pat (1–2 %) <i>SNURF</i> :TSS-DMR, LOM (1 %) <i>UBE3A</i> point mutations (10 %)	familial structural variants possible familial structural variants possible 0–15 % due to an IC deletion autosomal-dominant inheritance with parental imprinting		Severe intellectual disability, microcephaly, no speech, unmotivated laughing, ataxia, seizures, scoliosis
Central precocious puberty 2 (CPPB2) 615356	Unknown	15q11.2	<i>MKRN3</i> point mutations	autosomal-dominant inheritance with parental imprinting		Early activation of the hypothalamic–pituitary–gonadal axis resulting in gonadotropin-dependent precocious puberty
Schaaf–Yang syndrome (SYS) 615547	Unknown	15q11.2	<i>MAGEL2</i> point mutations	autosomal-dominant inheritance with parental imprinting		Delayed psychomotor development, intellectual disability, hypotonia
Pseudohypoparathyroidism 1B (PHP1B) 603233	Unknown	20q13	del(20q13)mat (single cases) <i>GNAS</i> DMRs, LOM (>60 %) upd(20)pat (2–20 %) <i>GNAS</i> point mutations	familial structural variants possible familial (structural) variants possible, maternal effect variants in case of MLID familial structural variants possible autosomal-dominant inheritance with parental imprinting	12.5 %	Resistance to PTH and other hormones, Albright hereditary osteodystrophy, subcutaneous ossifications, feeding behaviour anomalies, abnormal growth patterns
Mulchandani–Bhoji–Conlin syndrome (MBCS) 617352	Unknown	20	upd(20)mat	familial structural variants possible		IUGR, PNGR, feeding difficulties