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technologies

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Clinical features of post-ART and non-ART children with BWS

The 25 post-ART cases were conceived by IVF (n¼12) or ICSI (n¼13). Molecular genetic analysis revealed that 24 of the 25 post-ART children had LOM at KvDMR1 (no molecular cause was found in one post-ART child conceived by IVF).

In view of the known genotype…phenotype correlations of BWS (see Cooperet al., 2005 and references within), we compared the

Loss of methylation at SNRPN DMR in a patient with Beckwith…Widemann syndrome.
Reverse strand Pyrosequencing trace. Percentage of methylated cytosines is represented as the percentage of guanine (G) and the percentages unmethylated cytosines which normally will be represented by thymine is represented by alanine (A) on the reverse strand. (A) Normal control, (B) patient with LOM at SNRPN

Clinical and molecular characteristics of Imprinting Centre 2 defect Beckwith…Wiedemann syndrome patients with additional loss of methylation at other imprinted loci

	Patient 1	Patient 2	Patient 3	Patient 4	Patient 5	Patient 6
Sex	M	F	M	F	M	M
ART	IVF	ICSI	ICSI	No	No	No
Pregnancy	Singleton	Singleton	Twin	Singleton	Singleton	Singleton
Macrosomia	No	NR	No	No	Yes	Yes
Exomphalos	Yes	No	No	Yes	Yes	No
Umbilical Hernia	No	Yes	Yes	No	No	Yes
Macroglossia	No	Yes	Yes	Yes	Yes	Yes
Hemihypertrophy	Yes	No	No	No	No	Yes
Embryonal Tumour	No	No	No	No	No	No
Ear creases	Yes	No	Yes	NR	Yes	Yes
Neonatal Hypoglycaemia	No	Yes	Yes	Yes	Yes	Yes
Facial Naevus Flammeus	Yes	Yes	Yes	No	Yes	Yes
6q24 (ZAC) methylation	Normal	Normal	LOM	Normal	Normal	LOM
MI N¼(0.65 … 1.78)	1.25	1.23	0.55	1.22	0.93	0.61
7q32 (PEG1) methylation	LOM	LOM	Normal	LOM	LOM	Normal
15q13 (SNRPN) methylation	Normal	LOM	Normal	Normal	Normal	Normal
MI N¼(0.55 … 1.1)	0.84	0.05	0.98	0.82	0.77	0.89
14q32 (DLK1) methylation	Normal	Normal	Normal	Normal	Normal	Normal
MI N¼(0.5 … 1.4)	0.91	0.82	0.83	0.85	0.76	0.88
11p15.5 KvDMR1 methylation	LOM	LOM	LOM	LOM	LOM	LOM
MI	0.04	0.0	0.0	0.12	0.0	0.02

M, male; F, female; IVF, in vitro fertilization; ICSI, intra-cytoplasmic sperm injection; LOM, loss of methylation, MI, methylation index, NR, not recorded, N, normal range.

BWS children with IC2 defects might also display loss of methylation at other non-11p15.5 imprinting region DMRs. We found significantly higher frequencies of loss of methylation at DMRs unlinked to 11p15.5 in ART cases than in non-ART cases. This contrasts with the results of Rossignol et al. (2006) who found similar rates in both groups, but could be consistent with the hypothesis that differences in phenotype between ART and non-ART IC2 defect BWS patients might be caused by epigenetic differences at non-11p15.5 loci.