Supplementary table 1: Material and method used in the methylation test.

	Reagents	Amount		Primer sequence	Size
	H_2O	9 µl			
	Master Mix 2X	12 µl (1X)			
	DMSO	1.25 µl (5%)			
Primers	SNRPN_F* (50uM)	312 nM		AGGTCATTCCGGTGAGGGAGG	490 bp
	SNRPN_R (50uM)	312 nM		ACCGCAGACACCCGCAATAGG	
	H19_F* (50uM)	250 nM		GGCAACATGCGGTCTTCAGAC	420 bp
	H19_R (50uM)	250 nM		TCCGGAGACAGGGCTGAGCA	
	MEG3_F* (50uM)	250 nM		TCCTGACATGTTGCAGTCTTG	386 bp
	MEG3_R (50uM)	250 nM		CTCCACAACACCCGAAGCCAC	
	KCNQ1OT1_F* (50uM)	187 nM		CCTGTCATTGGCCGAAAGAGTC	366 bp
	KCNQ1OT1_R (50uM)	187 nM		GTTCGAGGGTCGCTGCAGC	
	HpaII[1] /H2O	10 U	1 µl		
	DNA	50 ng			
	TOTAL	25 µl			
		Digested	Undigested		

PCR program	
37ºC X 60 min	
94ºC X 1 min	
96ºC X 40 s	X 28 cycles
59ºC X 1 min	
72ºC X 1 min	
72ºC X 7 min	

* labelled with FAM

[1] New Englad Biolabs

F: forward; R: reverse

Supplementary Table 2: Segregation microsatellite markers used to evaluate a uniparental disomy as the cause of the methylation alteration. PCR reagents and programs, as well as sequence and size of each amplicon used in the PCR reactions. All reagents are from Roche except from Taq polymerase that is from

Marker (location)	Sequence	PCR reagents	PCR program	Size (bp)	Chr
D11S1984 (1.57Mb)	GGGTGACAGAGCAAAATTCT ACACCTGGATCTTGGACTCA	1µl gDNA (100ng/µl) 1X PCR reaction Buffer 2.5U Taq polimerase 480µM dNTPs 1µM Primer F 1µM Primer R 25µl V_f	3min 94ºC 30s 94ºC 30s 60ºC-0.5ºC/cy (10 cycles) 30s 72ºC 30s 94ºC 30s 55ºC (20 cycles) 30s 72ºC 7min 72ºC	170-202	11
D11S2362 (4.91Mb)	TGGACTATAGGACCCCCTTC GAGAACAGCCTGTCACACCT			209-230	
D11S1999 (10.72Mb)	TACATGGCAGCAGGCATATA GAGTAAACAAGATTGCTAGATAGGC			109-137	
D14S288 (44.10Mb)	AGCTAGACTCTGCCATAAACA TGGAGACAGGAACAACACAC			189-209	14
D14S68 (88.63Mb)	GAGAGGTGGTTTTCAGTGGT TCAGGGATAGTTGGTGGGTA			148-172	
D14S1006 (101.18Mb)	TTCCACAGGGCAAGCAGTA TTCTGGCAAAACCCAACC			121-155	
D14S985 (101.30Mb)	CAGTGTGACCTTAAACAAGTCG CCTGTGGGGTAGATACACGA			118-142	
D14S72[1] (21.37Mb)	TGTAAAGTTTTGTACATGGTGTAAT TCCTAACATTCTGCTACCCA			257-271	
D14S1023[1] (21.44Mb)	TGCATTTCCCGTAGACATT GACTCTTGTAGTTCTTTGAAGCC			93-109	
D14S990[1] (23.59Mb)	GTCCACTTGGTCATGGAAAC AAGTTGCACTGTGACTGGG			135-161	
D15S1035 (22.91Mb)	CACCCCCATGCAGAGTGAG CCAAAGGCCAAGACCTGCC			175-265	15
D15S11 (24.09Mb)	GACATGAACAGAGGTAAATTGGTGG GCTCTCTAAGATCACTGGATAGG			243	
D15S122 (25.68Mb)	GATAATCATGCCCCCCA CCCAGTATCTGGCACGTA			143-159	
GABRB3 (26.74Mb)	CTCTTGTTCCTGTTGCTTTCAATACAC CACTGTGCTAGTAGATTCAGCTC			180-200	

[1] Markers used to confirm duplication of patient 5 and the paternal origin of the alteration.

F: forward; R: reverse; V_f: final volumen