Table 1 Genotype–phenotype correlations in six cases of unstable ring chromosome 15

Case	Karyotype	Molecular karyotype	Igf1r	Stature
Case 1 (C.C.)	46,XX,r(15)(p11.2,q26)[88]/45,XX,-15[8]/ 47,XXr(15)(p11.2q26)x2[4]	2.1 Mb deleted (last oligonucleotide present 97,956 Mb; first oligo deleted 98.029 Mb)	Not deleted	At 25°
Case 2 (A.M.)*	46,XY,r(15)(p11.2,q26)[80]/45,XY,-15[15]/46,XY,dic r(15)(p11.2q26p11.2q26)[5]	1.6 Mb deleted (last BAC present RP11-90E15; first BAC deleted RP11-118O18)	Not deleted	At 25°
Case 3 (P.K.)	46,XX,r(15)(p11.2,q26)[80]/45,XX,-15[18]/ 47,XXr(15)(p11.2q26)x2[2]	4.9 Mb deleted (last oligonucleotide present 95,128 Mb; first oligo deleted 95.258 Mb)	Deleted	<3 rd
Case 4 (T.S)*	46,XY,r(15)(p11.2,q26)[91]/45,XY,-15[9]	4.8 Mb deleted (last BAC present RP11-667G9; first BAC deleted RP11-14A1)	Deleted	<3 rd
Case 5 (B.T.)	46,XY,r(15)(p11.2,q26)[86]/45,XY,-15[10]/46,XY,dic r(15)(p11.2q26p11.2q26)[4]	4.6 Mb deleted (last oligonucleotide present 95,258 Mb; first oligo deleted 95.523 Mb)	Deleted	<3 rd
Case 6 (M.M.)	46,XY,r(15)(p11.2,q26)[97]/45,XY,-15[3]	3.8 Mb deleted (last oligonucleotide present 95,523 Mb; first oligo deleted 96.305 Mb)	Deleted	<3 rd

*These cases have been defined by fluorescence in situ hybridisation (FISH) with contiguous bacterial artificial chromosomes (BACs) covering the last 2.5 Mb of 15q (from 97.715 Mb to 99.00 Mb; UCSC http://genome.ucsc.edu/cgi-bin/hgGateway, genome assembly May 2004).

largely overlapping those of distal 22q deletion. Moreover, recent papers have demonstrated that intact ring chromosomes may cause areas of hypopigmentation along the lines of Blaschko as the only sign of ring induced mosaicism,[14] or specific features such as a characteristic type of epilepsy and electroencephalographic pattern as reported for several ring (20) chromosomes,[15] thus weakening the hypothesis of the "ring syndrome".

We think that present data, showing that extreme short stature in ring chromosomes 15 associates with the haploinsufficiency of IGF1R rather than ring instability, further weakens the concept of the ring chromosome syndrome phenotype. Moreover, in all the array experiments we made, the ring deletion region has a log ratio between −0.8 and −1.2, clearly indicating a non-mosaic situation. Although this is likely to be due to the low level of mosaicism that makes impossible to detect this cell line with this technology, we cannot exclude the possibility that the ring instability is not present in blood DNA and appears only in cultured cells, being an in vitro phenomenon rather than an in vivo one.

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CORRECTION

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There was an error in an article published in the August issue of the journal (Hagerman PJ. The fragile X prevalence paradox. *J Med Genet* 2008;**45**:498–9). The sentence "Using an aggregate value (189/239,793; 1/126)…" which appeared on page 1, column 3, main paragraph, should read as follows: "Using an aggregate value (189/23,793; 1/126)…".