

Table S2 Cases with multiple sSMCs without apparent clinical abnormalities.

Table S2A Postnatal cases.

Phenotype	sSMCs	Coordinates ¹ (Mb, hg19)	Most likely explanation of normal phenotype	Reference [sSMC database ²]
42 year-old phenotypically normal woman, karyotyped because of inv dup(14/11) in child	inv dup(14/22)(q11.1) inv dup(15)(q11.1)	n.d. n.d.	does not contain euchromatin does not contain euchromatin	Gloning (Munich), unpubl. [mult 2-29]
28 year-old phenotypically normal woman, karyotyped because of recurrent abortion	min(6):(p11.2→q12:) min(20):(p11.21→q11.1:)	57.15 - 63.30 24.73 - q11.1	defines uncritical region of chrom.6 defines uncritical region of chrom.20	Guediche et al., 2012 [113] [mult 2-39]
31 year-old, phenotypically normal woman, karyotyped because of infertility	der(1):(p11.1→q12:) der(7):(p11.1→q11.21:)	n.d. n.d.	does not contain euchromatin within uncritical region of chrom.7	Wagner and Stibbe (Hannover), unpubl. [mult 2-45]
normal woman, no additional information	min(2):(p11.1→q11.1:) idic(18):(p11.21→q11.1::q11.1→p11.21:) mar3(?)	n.d. 0.516 Mb of 18p ?	does not contain euchromatin within uncritical region of chrom.18 ?	Castronovo et al. 2013 [87] (case 15) [mult 3-11]
normal adult male, no additional information	min(3):(p11.1→q11.1:) r(6)::(p11.2→q11.1::) r(9)::(p11.2→q12::) min(13)(pter→q12:)	n.d. n.d. n.d. n.d.	does not contain euchromatin within uncritical region of chrom.6 within uncritical region of chrom.9 probably within uncritical region ² of chrom.13	M. Vejsic (Belgrade) unpublished [mult 4-8]

Table S2B Prenatal cases.

Phenotype	sSMCs	Coordinates ¹ (Mb, hg19)	Most likely explanation of normal phenotype	Reference [sSMC database ²]
amniocentesis due to advanced maternal age; normal male baby at birth	r(3) r(?) but not 2,8,13/21,14/22,15,18,20,X,Y	n.d. n.d.	probably within uncritical region of chrom.3 probably within uncritical region ~16% of cells in blood do not have sSMCs	Viersbach et al. 1998 [23] (case 28) [mult 2-14]
amniocentesis due to advanced maternal age; at 8 years growth delay, but otherwise normal girl	r(X)(:p11.21→q12:) inv dup(15)(:q11.1→q11.2:)	57.73 - 64.59 0.00 - 21.05	within uncritical region of X chrom. within uncritical region of chrom.15 ~20% of amniotic cells do not have sSMCs	A. Dufke, (Tübingen), unpublished [mult 2-17]
amniocentesis due to positive serum screening, normal ultrasound findings, normal female baby at 3 months of age	mar(13/21) mar(non-acrocentric)	n.d. n.d.	probably does not contain euchromatin probably does not contain euchromatin	Huang et al. 2006 [126] (case 106) [mult 2-23]
amniocentesis due to positive serum screening, normal baby at 9 months	mar(14/22) in ~6% of cells mar(20) in ~6% of cells	n.d. n.d.	probably does not contain euchromatin probably within uncritical region of chrom.20 94% of metaphases do not have the sSMCs	Baldwin et al. 2008 [13] (case 23) [mult 2-43]
amniocentesis due to slight ultrasound anomalies; at birth overgrowth, at 1 year normal development	der(10)(p11.22→p11.1) der(12)(p11.21→q12)	34.3 - 38.7 32.0 - 40.7	almost completely in uncritical region chrom.10 within uncritical region of chrom.12	T. Liehr (Jena), unpublished, case provided by family [mult 2-46]
amniocentesis due to advanced maternal age; normal ultrasound, normal male baby at 20 months	mar(4) mar(8) mar(non-acrocentric)	n.d. n.d. n.d.	probably within uncritical region of chrom.4 probably within uncritical region of chrom.8 probably does not contain euchromatin	Huang et al. 2006 [126] (case 104) [mult 3-3]

Table S2B – Continued - Prenatal cases.

Phenotype	sSMCs	Coordinates ¹ (Mb, hg19)	Most likely explanation of normal phenotype	Reference [sSMC database ²]
amniocentesis due to possible bladder outlet obstruction seen at ultrasound; male baby; Apgar 9/10/10, mild macrocephaly; unilateral hydro-nephrosis; at 4 months normal development	?r(1) in ~84% of cells	n.d.	probably within uncritical region of chrom.1	Ulmer et al. 1997 [114] [mult 7-1]
	?r(3) in ~90% of cells	n.d.	probably within uncritical region of chrom.3	
	r(11) in ~80% of cells	n.d.	probably within uncritical region of chrom.11	
	min(14) in ~88% of cells	n.d.	probably within uncritical region of chrom.14	
	min(20) in ~74% of cells	n.d.	probably within uncritical region of chrom.20	
	min(21) in ~94% of cells	n.d.	probably within uncritical region of chrom.21	
min(X) in ~83% of cells	n.d.	probably within uncritical region of X chrom.		
amniocentesis due to advanced maternal age; normal ultrasound findings; at 2 years normal boy except for hypospadias and undescended testes	der(1/5/19) in 100% of amniocytes	n.d.	probably within uncritical region chrom.1/5/19	Chen et al. 2006 [115] [mult 7-2]
	der(2) in ~16% of amniocytes	n.d.	probably within uncritical region of chrom.2	
	der(4) in ~16% of amniocytes	n.d.	probably within uncritical region of chrom.4	
	der(6) in 100% of amniocytes	n.d.	probably within uncritical region of chrom.6	
	der(9) in 100% of amniocytes	n.d.	probably within uncritical region of chrom.9	
	der(10) in ~5% of amniocytes	n.d.	probably within uncritical region of chrom.10	
der(13/21) in ~77% of amniocytes	n.d.	probably does not contain euchromatin		

Notes

¹ if available, coordinates are given in megabases (Mb), according to hg19; n.d. not determined;

² see <http://ssmc-tl.com/sSMC.html> [14] for demarcation of the pericentromeric regions that do not cause a phenotype when present in three copies.