

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	Glöning (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of chrom.6 within uncritical region of chrom.9 probably within uncritical region ² of chrom.13	M. Vejsic (Belgrade) unpublished [mult 4-8]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 within uncritical region of chrom.15 ~20% of amniotic cells do not have sSMCs	A. Dufke, (Tübingen), unpublished [mult 2-17]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 macro-, cephalic; unilateral hydro-nephrosis; at 4 months normal development	?r(1) in ~84% of cells ?r(3) in ~90% of cells r(11) in ~80% of cells min(14) in ~88% of cells min(20) in ~74% of cells min(21) in ~94% of cells min(X) in ~83% of cells	n.d. n.d. n.d. n.d. n.d. n.d. n.d.	probably within uncritical region of chrom.1 probably within uncritical region of chrom.3 probably within uncritical region of chrom.11 probably within uncritical region of chrom.14 probably within uncritical region of chrom.20 probably within uncritical region of chrom.21 probably within uncritical region of X chrom.	Ulmer et al. 1997 [114] [mult 7-1]
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 der(2) in ~16% of amniocytes der(4) in ~16% of amniocytes der(6) in 100% of amniocytes der(9) in 100% of amniocytes der(10) in ~5% of amniocytes der(13/21) in ~77% of amniocytes	n.d. n.d. n.d. n.d. n.d. n.d. n.d.	probably within uncritical region chrom.1/5/19 probably within uncritical region of chrom.2 probably within uncritical region of chrom.4 probably within uncritical region of chrom.6 probably within uncritical region of chrom.9 probably within uncritical region of chrom.10 probably does not contain euchromatin	Chen et al. 2006 [115] [mult 7-2]

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.