

# GM00321 Fibroblast from Skin, Arm

**Description:**

XXX AND XXXXY SYNDROME  
ANEUPLOID CHROMOSOME NUMBER - NON-TRISOMIC

**Affected:**

No

**Gender:**

Female

**Age:**

40 YR (At Sampling)

<b>Repository</b>	NIGMS Human Genetic Cell Repository
<b>Subcollection</b>	Apparently Healthy Collection
<b>Biopsy Source</b>	Arm
<b>Cell Type</b>	Fibroblast
<b>Tissue Type</b>	Skin
<b>Transformant</b>	Untransformed
<b>Race</b>	Caucasian
<b>Ethnicity</b>	Sardinian
<b>Country of Origin</b>	USA
<b>Family Member</b>	2
<b>Relation to Proband</b>	mother
<b>Confirmation</b>	Karyotypic analysis and Case history
<b>Species</b>	Homo sapiens
<b>Common Name</b>	Human
<b>Remarks</b>	Unaffected mother of affected child (GM00326) and unaffected child (GM00323); normal RBC G6PD level; Xg(a) antigen positive; initial karyotype analysis reveals subject is 46,XX with random chromosome losses seen in 12% of cells, random chromosome gains seen in 2% of cells, and random chromosome aberrations seen in 2% of cells;
<b>PDL at Freeze</b>	8.8
<b>Passage Frozen</b>	6

<b>IDENTIFICATION OF SPECIES OF ORIGIN</b>	Species of Origin Confirmed by Chromosome Analysis
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<b>Remark</b>	Unaffected mother of affected child (GM00326) and unaffected child (GM00323); normal RBC G6PD level; Xg(a) antigen positive; initial karyotype analysis reveals subject is 46,XX with random chromosome losses seen in 12% of cells, random chromosome gains seen in 2% of cells, and random chromosome aberrations seen in 2% of cells;
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<b>Visentini S, De Nuccio C, Bernardo A, Pepponi R, Ferrante A, Minghetti L, Popoli P</b> , The stimulation of adenosine A2A receptors ameliorates the pathological phenotype of fibroblasts from Niemann-Pick type C patients The Journal of neuroscience : the official journal of the Society for Neuroscience33:15388-93 2013
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<b>PubMed ID:</b> <a href="#">24068806</a>
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<b>PubMed ID:</b> <a href="#">21177337</a>
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dbSNP	<a href="#">dbSNP ID: 16730</a>
GEO	<a href="#">GEO Accession No: GSM520733</a>
	<a href="#">GEO Accession No: GSM520734</a>
	<a href="#">GEO Accession No: GSM520735</a>
	<a href="#">GEO Accession No: GSM520736</a>
	<a href="#">GEO Accession No: GSM520737</a>
	<a href="#">GEO Accession No: GSM520738</a>
	<a href="#">GEO Accession No: GSM608313</a>
	<a href="#">GEO Accession No: GSM608314</a>
View	<a href="#">pedigree</a>
Cumulative PDL at Freeze	16
Passage Frozen	6
Split Ratio	1:4
Temperature	37 C
Percent CO2	5%
Percent O2	AMBIENT
Medium	Eagle's Minimum Essential Medium with Earle's salts and non-essential amino acids
Serum	15% fetal bovine serum Not inactivated
Substrate	None specified

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