

GM00326 Fibroblast

Description:

XXXY AND XXXXY SYNDROME
ANEUPLOID CHROMOSOME NUMBER - NON-TRISOMIC

Affected:

No Data

Gender:

Male

Age:

6 YR (At Sampling)

Repository	NIGMS Human Genetic Cell Repository
Subcollection	Chromosome Abnormalities
Cell Type	Fibroblast
Transformant	Untransformed
Race	Caucasian
Country of Origin	USA
Family Member	4
Relation to Proband	proband
Confirmation	Karyotypic analysis after cell line submission to CCR
ISCN	49,XXXXY
Species	Homo sapiens
Common Name	Human
Remarks	Normal RBC G6PD level; Xg(a) antigen positive; severe retardation; hypogonadism; mother is GM00321 Fibroblast
Passage Frozen	11
IDENTIFICATION OF SPECIES OF ORIGIN	Species of Origin Confirmed by Nucleoside Phosphorylase, Glucose-6-Phosphate Dehydrogenase, and Lactate Dehydrogenase Isoenzyme Electrophoresis and by Chromosome Analysis
Cytogenetics	Chromosome X: ANEUPLOID Aneuploid Segment (+)Xpter>Xqter
Remark	Normal RBC G6PD level; Xg(a) antigen positive; severe retardation; hypogonadism; mother is GM00321 Fibroblast
Treff NR, Su J, Tao X, Northrop LE, Scott RT	Single-cell whole-genome amplification technique impacts the accuracy of SNP microarray-based genotyping and copy number analyses Molecular human reproduction17:335-43 2010
PubMed ID:	21177337
Bonifas JM, Morley BJ, Oakey RE, Kan YW, Epstein EH Jr	Cloning of a cDNA for steroid sulfatase: frequent occurrence of gene deletions in patients with recessive X chromosome-linked ichthyosis. Proc Natl Acad Sci U S A84:9248-51 1987
PubMed ID:	3480541
Wolf SF, Marenzi CE, Migeon BR	Isolation and characterization of cloned DNA sequences that hybridize to the human X chromosome. Cell21:95-102 1980
PubMed ID:	7190879
dbSNP	dbSNP ID: 20818
View	pedigree
Passage Frozen	11

Split Ratio	1:3
Temperature	37 C
Percent CO2	5%
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
Subcultivation Method	trypsin-EDTA

Pricing

Commercial/For-profit:

\$130.00 USD

Academic/Non-profit/Government:

\$87.00 USD