

MOSAICISM COMPATIBLE WITH LIFE (I)

XX+21/XX MOSAICISM

Paternal age 40, maternal age 36

* AF sample in another lab XX+21/XX, then sent to our lab for further studies

* Referred to our lab for additional studies: CVS+AF+cord blood

Direct FISH on cord blood and CVS 6% and 22% of trisomy 21 respectively

Interruption of pregnancy

30 cells_ cytogenetic analysis of the cultured AF did not reveal the presence of the trisomy 21 cell line

Importance of the direct studies since mosaics can be lost in culture



45,X/47,XX+18/46,XX

Maternal age 39 + 46,XX,t(1;8)(q44;q24)

Paternal age 41

AF revealed only the presence of the 45,X [15] cell line. Went on with pregnancy.

At birth she presented Turner Syndrome features. Cytogenetic analysis showed a
mos 45,X[5]/47,XX+18[35]/46,XX[3]

Karyotype at the age of 15 years 45,X[9]/47,XX,+18[21]

She only shows Turner Syndrome features with a normal intelligence

Complexity of the mosaics

MOSAICISM COMPATIBLE WITH LIFE (II)

XX+mar/XX MOSAICISM (Referred sample)

Maternal age 37

AF sample- subculture 25% XX+mar

Original XX+mar/XX

Parental karyotypes normal

Origin of marker chr. 13;21 by FISH. No apparent presence of euchromatin

Reported: low risk of phenotypic manifestations according to the literature. Went on with pregnancy.

Today she is 2 and a half years old.

She has mild speech difficulties, goes to the speech therapist.

No comprehension difficulties.

Hair on her chest and genitalia.

Further clinical evaluation required.

Array-CGH? More sophisticated techniques



NEW STRATEGY

