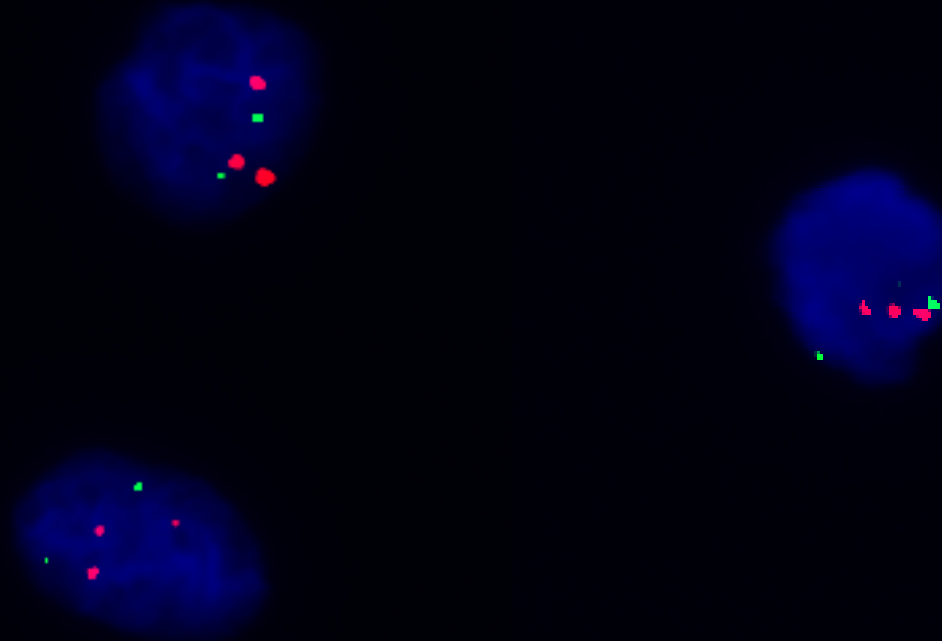


Referral

- Increased nuchal and scan
- Amniocentesis taken at 18 weeks
- Fetus has abnormal facial profile, retrognathia, hypoplastic nose, cleft palate, ventriculomegaly, hypoplastic left heart, pyelektasis, shortened humeri.

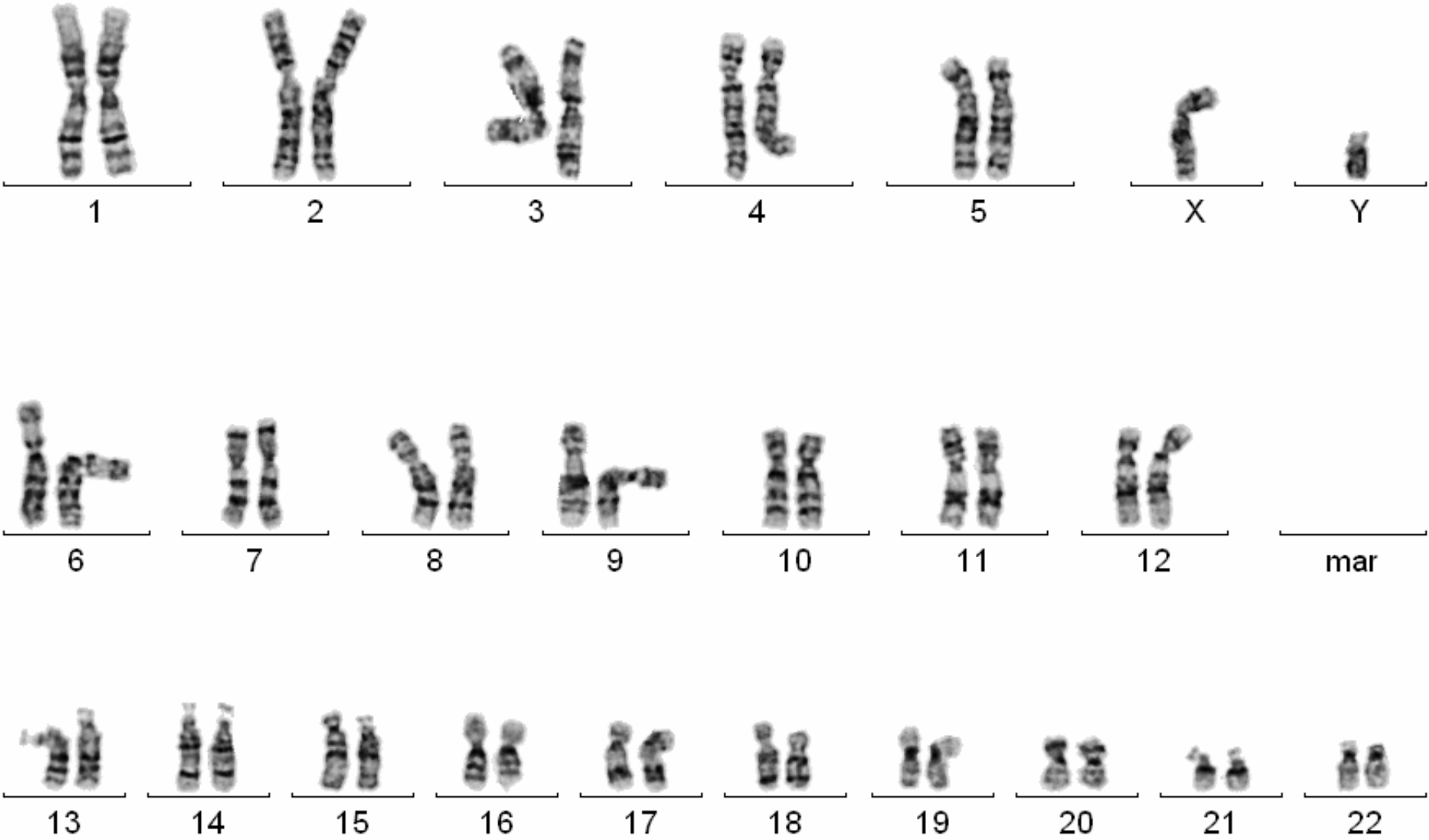
Aneuploidy FISH on uncultured amniocytes



Outcome of results

- Patient refused termination of pregnancy
- Brother in law had Down syndrome and therefore wasn't worried about an Down syndrome child
- Continued with pregnancy
- But culture came through.....

Culture - ?normal



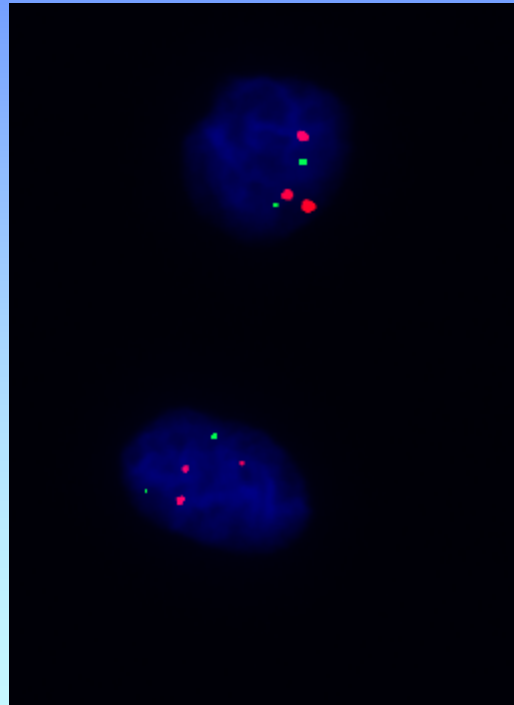
How could this occur?

- Mix up of samples
- Internal Audit suggested not
- Mosaic?
- Other

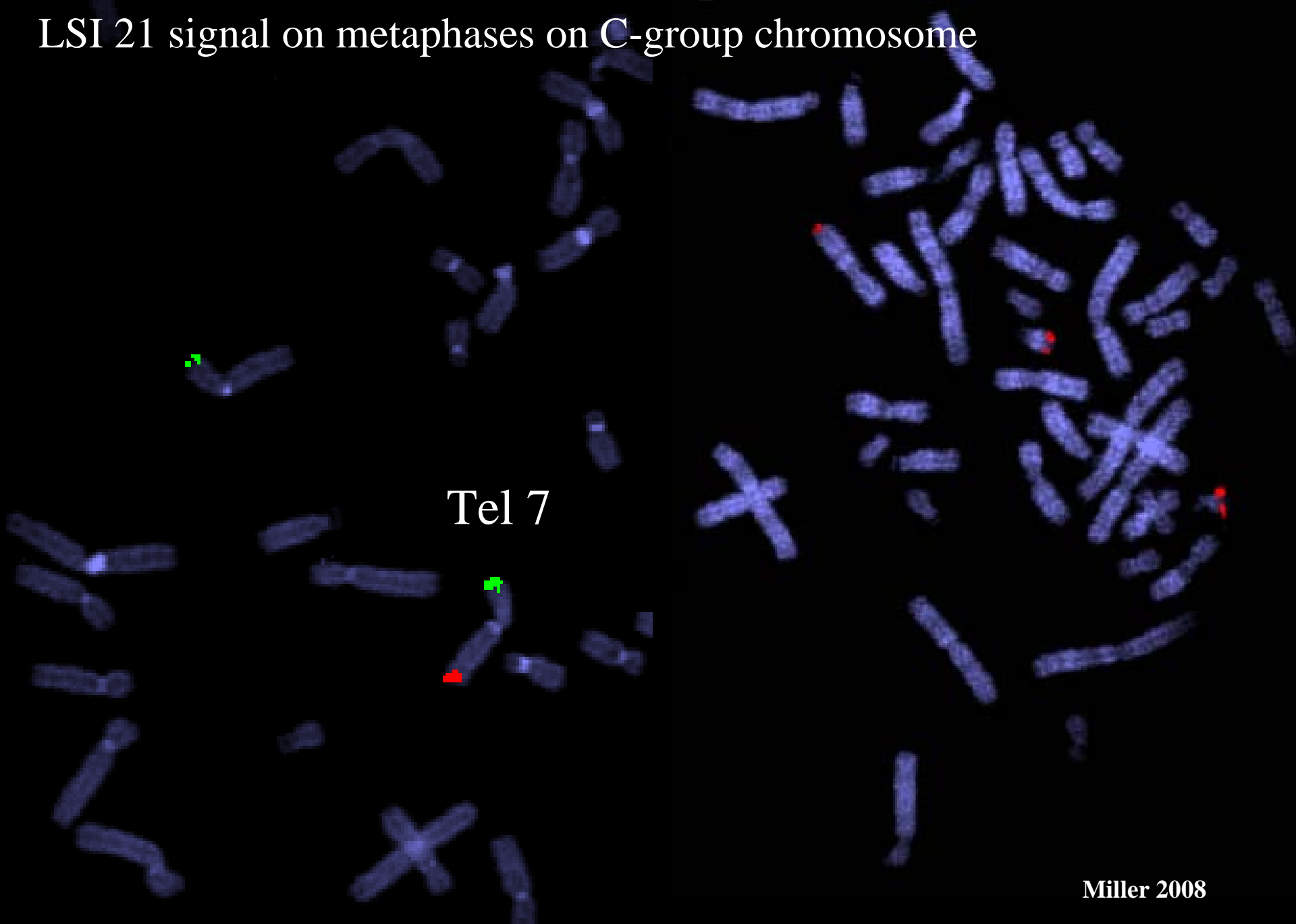
Decided to :

-FISH on cultured cells with LSI 21

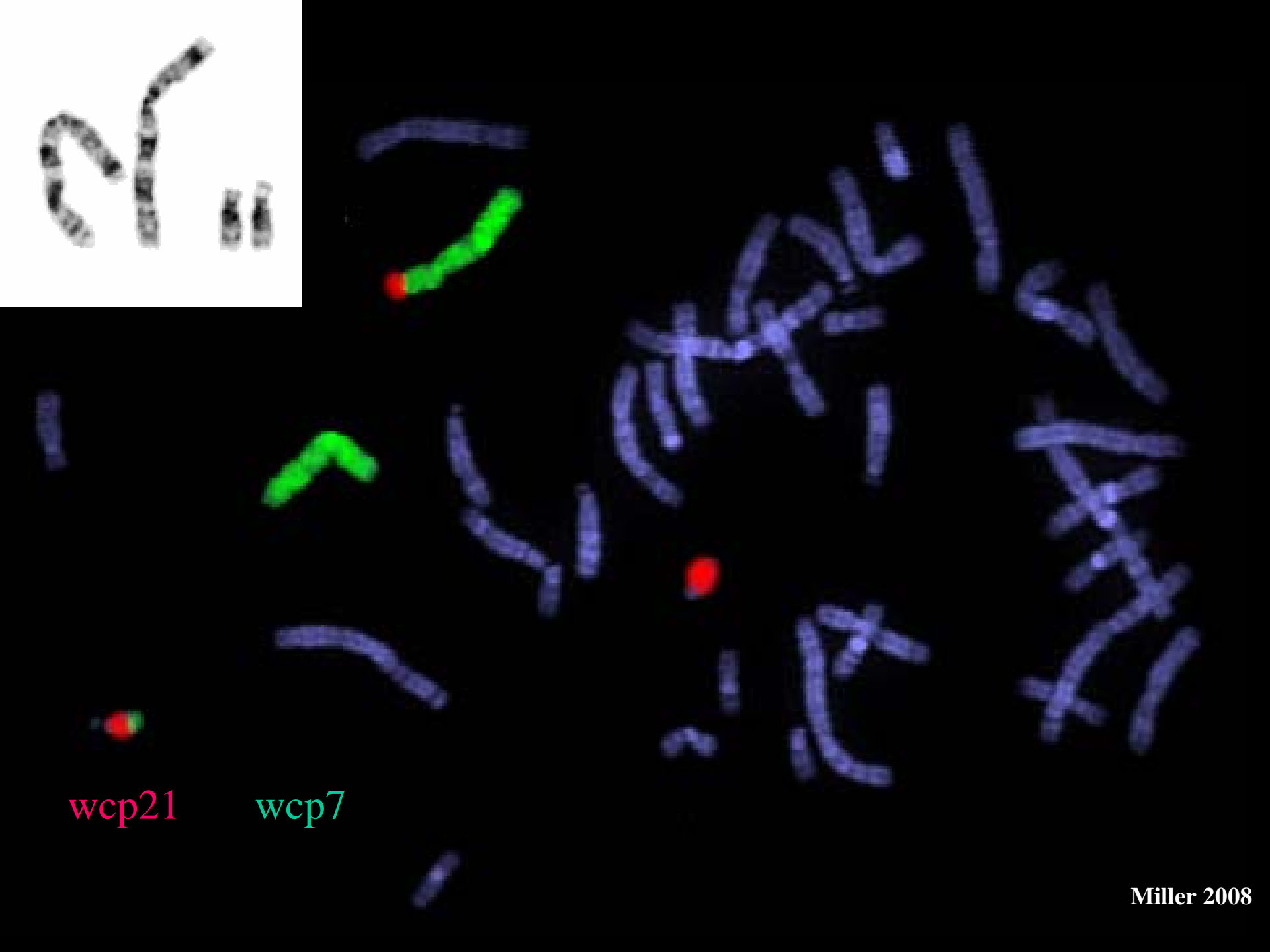
Cultured cells 3x 21 and seen on C-group chromosome in metaphases



LSI 21 signal on metaphases on C-group chromosome



Tel 7



wcp21

wcp7

Summary



- $t(7;21)(q34;q21)$ in dad
- Therefore $der(7)t(7;21)(q34;q21)$ in the fetus
- So what was the brother in laws' karyotype?
- Down's syndrome – not confirmed cytogenetically
– done after parents received genetic counselling
- Brother in law - $47,XY, t(7;21)(q34;q21) + 21$
- Different from fetus
- Brother in law therefore has a interchange trisomy or independent non-disjunction event
- Parents elected for a termination of pregnancy.