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On the significance of true trisomy 20 mosaicism in amniotic fluid culture

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Summary. Nine new cases of prenatally detected true mosaic trisomy 20 (T20) are reported. In three instances the fetuses were aborted. One fetus showed multiple malformations associated with a high percentage of T20 cells among amniotic fluid (AF) cells and fibroblasts of different fetal tissues. In two other fetuses only a slight facial dysmorphy was seen which was accompanied by a low percentage of T20 cells among AF cells. In five instances the pregnancies were carried to term, and normal somatic and psychomotor development of the children has been observed, in one case up to the age of 24 months. In one case the pregnancy is continuing. The T20 cells were not detected among cultured lymphocytes of these children.

A review of the hitherto known cases of prenatally detected mosaic T20 indicates a relationship between the prenatal findings and the fetal development. This may serve as a provisory basis for genetic counselling: in the case of a percentage above 50% of T20 cells among AF cells there seems to be a risk of about 50% for the fetus to be affected by severe anomalies. However, in cases of a prenatally detected mosaic T20 with a percentage equal to or less than 50, fetal or congenital malformations have not been observed among 23 individuals so far examined.

Introduction

In prenatal chromosome analysis a "true" mosaic is present if two different karyotypes were detected in at least two different clones or cultures. A mosaic with trisomy 20 (T20) has rarely been found among spontaneous abortions and newborns. However, this aberration is frequently detected in amniotic fluid cultures (Jacobs et al. 1974; Creasy et al. 1976; Kajii et al. 1980). The existence of this mosaic trisomy in amniotic fluid (AF) cells has recently been substantiated by biochemical evidence indicating a triplex gene dosage of adenosine deaminase in trisomic clones (Steinbach et al. 1985). The significance of a prenatally detected T20 mosaic, however, is still in doubt. This makes genetic counselling difficult in this case.

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A malformed fetus with T20 mosaic was reported by Boué et al. (1979), whereas several fetuses were reported to be apparently normal (e.g., Nevin et al. 1979; McDermott et al. 1981). Children born after prenatal diagnosis of T20 mosaicism were also normal (e.g., Kardon et al. 1979; Mascarello et al. 1980).

Case reports

Case 1. A healthy 40-year-old woman was referred for amniocentesis at 16 weeks in her sixth pregnancy. She had two previous miscarriages (M III, M IV). Amniotic fluid cultures were harvested on day 14. Chromosome analysis from two primary cultures revealed 11% T20 cells identified by G- and Q-banding. Amniocentesis was repeated at 20 weeks of gestation and two further primary cultures were separately harvested ten days later. Chromosome analysis on both cultures revealed 10% T20 cells (Table 2). The parents elected to terminate the pregnancy.

The fetus was 22 weeks old and female. Weight was 408 g and crown-heel length 27 cm. There was a slight craniofacial dysmorphy including a narrow skull with high forehead, hypertelorism, bilateral epicanthic folds, slightly "antimongoloid" slanting of palpebral fissures, and a distinct micro-retrogenia. The fetus also showed large and poorly differentiated auricles, a slight hypoplasia of the thenar eminences, an incomplete lobulation of the right lung, and a distinct meandering of the left ureter. Fetal cells cultured from rib, amnion, pericardium, diaphragm, and Achilles tendon revealed no evidence of T20 in a total of 259 metaphases analyzed from these tissues (Tables 3 and 4). The parents had normal karyotypes.

Case 2. A healthy 37-year-old woman was referred for amniocentesis at 16 weeks in her second pregnancy. The family history was unremarkable. Amniotic fluid cultures were harvested on day 16. T20 was detected in 93% of the cells from one primary culture and in 75% of the cells from a first subculture (Table 2). The pregnancy was terminated.

The 21-week-old fetus was female. Weight was 330 g and crown-heel length 24 cm. The following stigmata were recorded: high forehead, hypotelorism, slight "antimongoloid" slanting of the eyelashes, short nose, prominent philtrum, small mouth, microgenia, low set ears with poorly differentiated auricles, short neck, camptodactyly, anal fistula, and bilat-

Table 1. Prenatal diagnoses of true mosaic trisomy 20

Reference	Case no.	Sex	Consequence	Origin of T20
Hsu et al. (1976, 1978)	1	F	Termination	Unknown
Laurence and Gregory (1976)	2	F	Termination	Unknown
Gregory personal communication (1983)				
Rudd et al. (1977)	3	M	Termination	Unknown
	4	F	Termination	Placenta?
Rodriguez et al. (1978);	5	F	Termination	Unknown
Gardner personal communication (1983)	6	F	Continuation	Unknown
Boué et al. (1979)	7		Termination	Fetus
Golbus et al. (1979);	8	F	Termination	Unknown
personal communication (1983)	9	M	Continuation	
Kardon et al. (1979)	10	F	Continuation	Unknown
	11	M	Continuation	Unknown
	12	F	Termination	Unknown
Mikkelsen personal communication (1983)	13		Termination	Fetus
Nevin et al. (1979)	14	F	Termination	Unknown
Mascarello et al. (1980)	15	F	Continuation	Unknown
Weise and Quent (1980)	16	F	Termination	Unknown
Weise and Bernoth personal communication (1983)	17	F	Termination	Unknown
McDermott et al. (1981);	18	F	Termination	Placenta, fetus
personal communication (1983)				
Nisani et al. (1981)	19	M	Continuation	Unknown
Schwinger and Rehder (1981);	20	M	Termination	Fetus
Schwinger personal communication (1983)	21	M	Termination	Fetus
Watt et al. (1981);	22[a]	F	Termination	Fetus
personal communication (1983)				
Bosze et al. (1982);	23	F	Continuation	Unknown
Bosze and Toth personal communication (1983)				
Miny personal communication (1983)	24	M	Termination	Unknown
Present Case 1	25	F	Termination	Unknown
Present Case 2	26	F	Termination	Unknown
Present Case 3	27	M	Continuation	Unknown
Present Case 4	28	M	Continuation	Unknown
Present Case 5	21	M	Termination	Fetus
Present Case 6	29	M	Continuation	Unknown
Present Case 7	30	FF	Continuation	Unknown
Present Case 8	31	M	Continuation	Unknown
Present Case 9	32	M	Continuation	Unknown

[a] Karyotype: 45,X/46,X,+20

eral pes planus. There were lobulation anomalies of lungs and liver, a partly intrahepatic gallbladder, a pancreatic heterotopy into the submucosa of the duodenum, an enlarged renal pelvis, meandering of the ureters, and a slightly bicornic uterus. The heart was elongated and showed transposition of arteries, an infundibular pulmonary stenosis, hypoplasia of right ventricle, and hypoplasia of tricuspid and bicuspid valve. The T20 cells were recovered among fetal fibroblasts cultured from fascia lata and rib (Table 3). The parents had normal karyotypes.

Case 3. A healthy 40-year-old woman came to amniocentesis at 18 weeks in her third pregnancy. There were two previous miscarriages (M III, M IV). One primary amniotic fluid culture with an unknown number of clones was harvested after 20 days. Among 23 cells 22 showed T20. A second amniocentesis at 23 weeks provided four additional cultures harvested after 14 days. Ten clones examined in situ showed a normal male karyotype. However, 7% of the metaphases from one trypsinised primary culture again showed the trisomy (Table 2). The pregnancy was continued.

At 39 weeks of gestation a phenotypically normal boy was delivered by cesarian section. The baby weighed 2880g, length was 49cm, occipito-frontal head circumference (OFC) 34cm. Apgar scores were 8, 9, and 10. Chromosome analysis on peripheral lymphocytes revealed a normal karyotype 46,XY. At 8 months weight was 7,550g (10 percentile) and length was 70cm (25–50 percentile). He walked alone at 10 months. At 1 year and 3 months he weighed 9,400g (10 percentile) and was 80cm tall (50 percentile). At 2 years and 6

Table 2. Cytogenetic findings in amniotic fluid culture

Case no.	Week of pregnancy	Days of cultivation	T20/no. of cells or clones	
1	16		6/ 70	(9%)
	20		0/135	
2				(2%)
3			40/100	(40%)
	at		0/ 24	
4	20	<26	9/ 45	(20%)
	at		15/ 85	(18%)
5	17	16	6/ 24	(25%)[a]
6	22	14	3/ 35	(9%)[a]
			3/ 37	(8%)
7	20	7	99/100?	(99%)[a]
	at		56/ 60	(93%)[a]
8	14	14	20/ 90	(22%)
9	27.5	13	7/ 40	(18%)
10	18	15	26/100	(26%)
11	16	21	18/ 92	(20%)
12	17	21	6/ 70	(9%)
	20		0/135	
13	15	21	13/110	(12%)
	19	15–20	16/100	(16%)
14	16	15	19/ 41	(46%)
15			25/ 34	(74%)
			11/ 43	(26%)
16	16	16	16/ 70	(23%)
17	17	18	20/ 56	(36%)
18	16	11	26/ 42	(62%)
	18		8/ 14	(57%)
20	17	13	17/ 28	(61%)[a]
21	17	17	11/ 76	(14%)
	22		12/119	(10%)
22	18		15/ 30	(50%)
23	18	20	12/ 39	(31%)
24	16	15–21	3/ 3	(100%)[a]
	19	14	4/ 6	(67%)[a]
25	16	14	6/ 58	(10%)
	20	10	9/ 86	(10%)
26	16	16	27/ 29	(93%)
27	18	20	22/ 23	(1 clone?)
	23	14	0/ 10[a]	
	23	14	9/143	(6%)
28	16	18	30/ 85	(35%)
29	17	13	2/ 4	(50%)[a]
30	16	12	7/ 12	(58%)
			5/ 17	(29%)
31	16	15	21/ 88	(24%)
32	16	13	8/ 62	(13%)

[a] Clones in situ
at: at termination

months a report from his mother substantiated normal development.

Case 4. A 43-year-old woman had an amniocentesis at 16 weeks in her fourth pregnancy. The first pregnancy had resulted in a spontaneous abortion. The family history was otherwise unremarkable. A total of 19 clones from three trypsinised primary cultures were examined on day 18, and 35% of the cells analysed showed T20 (Table 2). The pregnancy was continued.

At term a phenotypically normal boy was delivered by cesarian section. Apgar scores were 9 and 10. Birth weight was 3930 g, length 54 cm, and OFC 36 cm. A chromosome analysis on peripheral lymphocytes revealed a normal male karyotype 46,XY. At 6 months (Fig. 2) his weight was 8950 g (75 percentile), length 72 cm (<90 percentile), and OFC 46 cm (96 percentile). At 8 months his psychomotor development was reported to be normal.

Case 5. This case has been briefly mentioned by Schwinger and Rehder (1981). The amniocentesis was performed in the 17th week of the first pregnancy of a healthy 33-year-old woman. Chromosomal analysis was carried out on three primary cultures harvested after 17 days and revealed 14% T20 mitoses (Table 2). The pregnancy was terminated.

Additional cultures were established from amniotic fluid punctured immediately before termination. Ten percent T20 cells were obtained from this material (Table 2). Fetal fibroblasts cultured from biopsies of rib, fascia lata, a metatarsal bone, umbilical cord, lung, and both ureters did not show a single T20 cell out of a total of 550 mitoses examined (Table 3). T20 cells were, however, detected among fibroblasts from both fetal kidneys, i.e., 4 out of 126 cells from the right and 1 out of 214 cells from the left kidney. Schwinger and Rehder (1981) reported 14% trisomic cells for the renal tissue of this fetus.

The fetus was male and 22 weeks old. Weight was 356 g and crown-heel length 25 cm. There was a small and high skull, hypertelorism, slight epicanthic folds, a prominent philtrum, and micro-retrogenia. The right hand showed a slight camptodactyly. The ureters were enlarged and showed meandering. There was macronesia of the pancreas (Schwinger and Rehder 1981).

Case 6. A healthy 38-year-old woman was referred for prenatal chromosome analysis at 17 weeks in her second pregnancy. The family history was unremarkable. Amniocyte clones were examined in situ after 13 days of culture. Two clones showed a normal 46,XY karyotype while two other clones consisted of T20 cells only (Table 2). The pregnancy was continued.

The baby was spontaneously born two days after term. Birth weight was 4000 g, length 55 cm, and OFC 36 cm. The newborn showed the following stigmata: Prominent occiput, impressed forehead, broad nose bridge, anteverted nostrils, narrow palpebral fissures with "mongoloid" slanting, slight epicanthus, deep-set auricles, paresis of the facial nerve on the right, short and broad neck with redundant skin posteriorly, laterally placed nipples, and a small omphalocele. There was a bilateral simian crease, an increased distance between the first and second toes, and a transverse crease on both plants. A chromosome analysis carried out on peripheral lymphocytes postnatally gave a normal male karyotype in 50 mitoses examined. The parents also showed normal karyotypes.

At 5 months and 3 weeks the baby's weight, length, and OFC corresponded to the 50 percentile and psychomotor development was normal for age. A neurologic examination was normal. Most of the stigmata recorded in the newborn were also recognised in the baby's parents or his elder brother. The only remaining deviation from the normal family trait

Table 3. Cytogenetic findings in fetal tissues after prenatal diagnosis of mosaic trisomy 20

Case no.	AF cells (% T20)[a]	T20 positive (% T20)	T20 negative (% T20)
7	93%	Kidney (90%)	Skin
26	93%	Fascia lata (40%), rib (18%)	
24	67%		Blood
20	61%	Kidney (6%)	Placenta, amniotic membrane, skin, renal pelvis, pericard, ureter, urinary bladder
18	57%	Placenta (100%), placental membrane (91%), skin (28%)	Lungs, amniotic membrane
22	50%	Cord (2%), skin (2%), lung (2%), ovary (2%)	Heart, liver
14	46%		Cord, placenta, skin
3	40%		Skin, lung, intestine liver, kidney, blood
5	25%		Fascia lata, skin
4	18%	Placenta (0.1%)[b]	
13	12%	Rectum[c]	Trachea, esophagus, fascia lata, Achilles tendon, ureters, amniotic membrane
21	10%	Kidney (14%)	Placenta, amniotic membrane, cord, skin, cord, skin, lung, metatarsal bone, renal pelvis, ureters, urinary bladder
25	10%		Amniotic membrane, rib, Achilles tendon, pericard, diaphragm
2	2%		Skin, cord blood

[a] Smallest positive value obtained if more than one experiment/amniocentesis
[b] Fetus not examined
[c] One cell; 503 cells examined in all tissues

Table 4. Phenotypic findings in fetuses after prenatal diagnosis of mosaic T20

Case no.	AF cells (% T20[a])/ fetal tissue (% T20)	Age (weeks)	Weight (g)	Length (CHL, cm)	Findings
7	93%/ 90%				Microcephaly, facial dysmorphy
26	93%/ 40%	21	330	24	Heart defect, lobulation anomaly of lung and liver, facial dysmorphy, camptodactyly, anal fistula
24	67%/		540	30.5	Facial dysmorphy, horseshoe kidney, cardiac anomalies
18	57%/ 28%				Normal fetus
20	61%/ 6%	20	394	27.5	Minor urinary tract anomalies, no functional disorder of kidney
22[b]	50%/ 2%				Fetal Turner syndrome
14	46%/ 0%	22	565		Slight facial dysmorphy
3	40%/				Unilateral hydronephrosis
17	36%/	23	480	31	Normal fetus
5	25%/ 0%				Normal fetus
16	23%/	23	400	30	Normal fetus
8	22%/	18	295	18[c]	Normal fetus
4	18%/	23	650	27[c]	Normal fetus
13	12%/>0%				Slight facial dysmorphy
21	10%/ 14%	22	356	25	Slight facial dysmorphy
25	10%/ 0%	22	408	27	Slight facial dysmorphy
1	9%/	20	370	27	No anomalies reported
12	9%/				No anomalies reported
2	2%/				Normal fetus

[a] See Table 3
[b] 45,X/46,X,+20; CHL: Crown-heel-length
[c] Crown-rump-length. Mean age: 21.4 weeks; mean weight: 425 g; mean length (CHL): 27.4 cm

Table 5. Findings in children born after prenatal diagnosis of mosaic T20

Case no.	AF cells (% T20[a])	Lympho-cytes (karyotype)	Fibro-blasts (karyotype)	Birth weight (g)	Birth length (cm)	Birth OFC (cm)	Clinical status (at age)
29	50%	46,XY		4000	55	36	Redundant skin of neck, small omphalocele (at birth); normal development (15 months)
28	35%	46,XY		3930	54	36	Normal (9 months)
23	31%	46,XX	46,XX	3450			Normal development (4 years)
30	29%	46,XX					Normal twins (3 months)
15	26%	46,XX					Normal
10	26%	46,XX	46,XX	3150			Normal
31	24%						Normal
11	20%	46,XY		3430			Normal
9	18%						Ascertained by intrauterine growth retardation; pregnancy continuing
32	13%						Pregnancy continuing
6	8%	46,XX	46,XX	3150			Normal (2 months)
27	6%	46,XY		2880	49	34	Normal (30 months)
19	4%	46,XY	46,XY				Normal

[a] See Table 3
Mean birth weight: 3445 g

was the redundance of skin on the neck and the small omphalocele. At 15 months the boy was again reported to be developing normally.

Case 7. A prenatal diagnosis of a mosaic trisomy 20 was made in a pregnancy with monoamniotic twins. The amniocentesis was performed at the 16th week. After 12 days there were 12 clones examined in situ. Seven clones had a female karyotype with trisomy of chromosome 20, five clones showed a normal 46,XX karyotype. A trypsinised culture gave five trisomic cells among 17 cells analyzed (Table 2). The chromosomes of the fetuses could not be distinguished.

An ultrasonic examination did not reveal any abnormal features. The pregnancy was continued and two girls were born normally. Chromosome analysis on lymphocytes from cord blood of both babies failed to detect a single trisomic cell among 50 metaphases analyzed for each individual. Both children were reported to be developing normally at the age of 3 months.

Case 8. A healthy 29-year-old woman was referred for amniocentesis at 16 weeks of her first pregnancy. Prenatal chromosome analysis was performed because she had had irradiation treatment for a lymphogranuloma. Amniotic fluid cultures were harvested after 15 days. Trisomy 20 metaphases were detected in three primary cultures using Q-banding. The total amount of T20 cells was 24% (Table 2).

A normal boy was born two days after term by cesarian section. Birth weight was 3300 g, Apgar scores were 9, 10, and 10. Chromosome analysis on cord blood did not reveal any trisomic cell among 68 lymphocytes analyzed.

Case 9. A healthy 39-year-old woman had an amniocentesis at 16 weeks of her fourth pregnancy. Two primary amniotic fluid cultures were harvested after 13 days. Metaphases with trisomy 20 were detected in both of them. There was a total of 13% trisomic cells identified by Q-banding (Table 2). The pregnancy is continuing.

Discussion

There have been 32 cases of prenatally detected mosaic T20 analysed. Data on reported cases were obtained from the literature and completed by personal communication with the investigators. In 19 instances the fetuses were aborted, whereas in 13 cases the pregnancies were continued.

We have no doubt that in the present cases the AF cells identified as T20 cells do contain an accessory chromosome 20. This has also been proven by recent biochemical evidence in the present Case 2 (Steinbach et al. 1985). The fetal origin of the T20 AF cells has been well established for many cases (Watt et al. 1981; McDermott et al. 1981). However, an extrafetal origin of T20 may also be possible (Kardon et al. 1979). Several examples are known in which a T20 cell line detected in amniotic fluid culture was not present in every fetal tissue studied (e.g., Boué et al. 1979). Evidence is accumulating that renal tissue is more often involved in T20 mosaicism previously detected in amniotic fluid (Schwinger and Rehder 1981), although Rudd et al. (1977) failed to detect T20 cells even in the kidney of his proband (Case 3, Table 3).

Abnormal somatic findings including deformations, malformations, and dysmorphic features were reported for a minority of T20 mosaic fetuses or newborns (Tables 4 and 5). Many individuals had discrete minor stigmata which may represent normal familial variants rather than anomalies indicating a discontinuous trait (e.g., present Case 6).

The incidence of true mosaic T20 in amniotic fluid cultures is about 1 per 2,000 as estimated from the experience with 10,000 amniocenteses carried out at the University Hospital Ulm. However, among all postnatally examined individuals there has been only one observation of a probable T20 mosaic (Wahlström et al. 1976). The cytogenetic documentation of another report by Carbonell et al. (1977) is not convincing, since the presented karyotype as well as the described phenotype suggest a mosaic trisomy 21 with a Gp+ variant rather than a mosaic T20.

The obvious discrepancy between the incidence of mosaic T20 in pre- and postnatally examined individuals may be explained by the following hypotheses: (1) The T20 mosaicism does not lead to a syndrome suggestive of a chromosomal imbalance. (2) An existing T20 mosaicism does not usually involve either lymphocytes or fibroblasts which are generally used for chromosome analysis.

The first hypothesis is substantiated by the fact that most fetuses and newborns examined after prenatal diagnosis of a mosaic T20 did not show severe abnormalities (Tables 4 and 5). In view of the possible limitation of the T20 cells to certain tissues in many cases, the clinical consequence, if any, may not be typical for a chromosome imbalance. The second hypothesis is substantiated (1) by the finding of normal karyotypes in lymphocytes from prenatally detected mosaic T20 individuals, (2) by the fact that the detection of T20 cells in fibroblasts cultured from skin biopsies of mosaic T20 fetuses is a rare event (Table 3), and (3) by the rarity of a mosaic T20 among spontaneous abortions (Creasy et al. 1976; Kajii et al. 1980).

A mosaic trisomy 20 obviously does not lead to either intrauterine growth retardation or specific malformations (Tables 4 and 5) and the significance of this prenatally detected mosaicism is not yet fully known. However, some guide lines may serve as a provisory basis for genetic counselling in the context of a prenatally detected T20 mosaicism.

There is a difference in the significance of the cytogenetic result between a mosaicism with a high percentage (above 50%) and a mosaicism with a low percentage (less than or equal to 50%) of T20 AF cells (Tables 4 and 5). In the first situation there is a good chance of detecting T20 cells in several fetal tissues and a risk as high as 60% or more for the child to be affected by severe anomalies involving brain, heart, or kidney (Table 4). In the second and most frequent situation (Table 6) there seems to be no good chance of detecting fetal T20 fibroblasts if the kidney is not included in the study (Table 3). The risk of the child being affected by a serious congenital anomaly seems to be very low (Tables 4 and 5): only five out of 23 informative individuals have been reported with some discrete stigmata not impairing normal development. However, there is still a lack of data on children with a prenatal diagnosis of mosaic T20 older than 4 years.

The first ("high risk") situation should only be assumed if the high percentage of T20 cells was confirmed after repeated amniocenteses (see Cases 23 and 25 in Table 2). The second ("low risk") situation, however, usually seems not to require a second analysis for substantiation.

The present data on cases with prenatally detected true T20 mosaicism suggest a difference between a "high risk" and a "low risk" situation. Such a difference might be due to the existence of two biologically different types of T20 mosaicism, e.g., one type due to meiotic nondisjunction with postzygotic loss of one chromosome 20 in some of the embryonic cells or to an early somatic mutation giving rise to T20 cells in all fetal tissues, and another type due to a mutation later in development leading to a limitation of aneuploid cells to some organs or even a single organ.

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Note added in proof

In the present case 9 the boy has been born normally. Birth weight was 3330 g, length 51 cm, OFC 34.5 cm. An intensive pediatric examination including kidneys, urinary tracts, brain and heart was normal.