POSTNATAL OUTCOMES FOLLOWING PRENATAL DIAGNOSIS OF MOSAICISM: a systematic review of literature

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OBJECTIVE. Prenatal detection of mosaicism (M) by amniocentesis or chronic villous sampling (CVS) is a challenge and counselling is often difficult, since only case reports are described in literature. The objective of this review was to pool these cases in order to obtain a large sample size of pregnancies affected with M.

METHODS. A search in PubMed, EMBASE, Medline, reference lists was made without limits of time. Key words were: aneuploidy, prenatal diagnosis, karyotype, amniocentesis, CVS, mosaicism. Inclusion criteria were: prenatal detection of M, placental and fetal karyotype obtained postnatally. Exclusion criteria were: postnatal diagnosis of M and non-English language publications. Postnatal outcomes were reviewed.

RESULTS. See table. In the placental M group, 3 (27%) fetuses were IUGR. In fetal&placental M, the only alive fetus had developmenta delay.

	FETAL M	PLACENTAL M	FETAL & PLACENTAL M
DIAGNOSIS			
CVS	1 (5%)	3 (27%)	2 (40%)
Amnio	18 (90%)	8 (73%)	3 (6%)
UCS	1 (5%)	0	0
MAJOR MALFORMATIONS	19 (95%)	1 (9%)	4 (80%)
SINGLE MALFORMATIONS	6 (30%)	1 (9%)	4 (80%)
MULTIPLE MALFORMATIONS	14 (70%)	0	0
TOP	14 (70%)	2 (18%)	4 (80%)
NEONATAL DEATH	1 (5%)	1 (9%)	0
ALIVE AND WELL	4 (20%)	8 (73%)	1 (20%)

CONCLUSION. Fetal mosaicism is associated with poor outcomes, whereas confined placental mosaicism has a better prognosis. Because of this discrepancy, prenatal diagnosis of mosaicism should prompt detailed ultrasound examination and fetal karyotype assessment in order to establish the origin of mosaicism.