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Postnatal outcomes following prenatal diagnosis of placental mosaicism: Case reports from literature

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Mosaicism is a rare genetic condition that occurs when two or more populations of cells are chromosomally different. The association between mosaicism and phenotypic abnormalities varies widely according to the degree of mosaicism and the genetic anomaly. Mosaic cells can affect fetal tissue, might be confined to the placenta or may involve both the fetus and placenta. Prenatal detection of mosaicism is obtained by amniocentesis or chronic villous sampling (CVS) and represents a clinical dilemma, since only case reports or very small series are described in literature. In addition, articles reporting about mosaicism focus on the genetic disease rather than the effects of mosaicism. We therefore performed a review of literature in order to pool case reports and small series and obtain a large sample size, which is essential for studying and counselling parents about a rare condition, such as mosaicism. For this purpose, we selected articles that were based on prenatal detection of mosaicism by invasive procedures and obtained placental and fetal karyotype postnatally or postmortem. Survival rates were defined as newborns alive and well within 28 days from birth. Perinatal outcomes were defined as presence of fetal malformation, intrauterine demise, termination of pregnancy, and postnatal examination or autopsy, and were stratified according to the characteristics of the mosaicism. From 33 articles, 36 cases of mosaicism were pooled. Of these, 20 (55.5%) were confined to fetal tissue, 11 (30.5%) to the placenta and 5 (14.0%) were both fetal and placental mosaicism. Fetal mosaicism was observed with amniocentesis (18 cases; 90.0%), CVS (1 case; 5.0%) or umbilical cord sampling (1 case; 5.0%) and was either structural (5 cases; 25.0%) or numerical (15 cases; 75.0%). All but one fetus presented major malformations. Six fetuses (30.0%) had single malformations and 14 fetuses (70.0%) presented multiple anomalies. Fourteen pregnancies (70.0%) were terminated, 1 pregnancy (5.0%) ended with fetal demise, and 1 neonatal death (5.0%) occurred because of prematurity, leaving 4 infants (20%) alive and well. Placental mosaicism was observed with amniocentesis (8 cases; 72.7%), or CVS (3 cases; 27.3%) and was either structural (1 case; 9.0%) or numerical (10 cases; 91.0%). Three fetuses (27.2%) were growth restricted. There were 1 fetus (9.0%) with transposition of great vessels and 3 (27.2%) fetuses with minor malformations. Two pregnancies (18.2%) were terminated, 1 (9.0%) neonatal death occurred for prematurity, and 8 (72.8%) liveborns were normal. In the 5 cases of both fetal and placental mosaicism, prenatal diagnosis was obtained by amniocentesis (3 cases; 60.0%) and CVS (2 cases; 40.0%). Four pregnancies (80.0%) were terminated because of major malformations and 1 fetus (20.0%) was apparently normal but developmental delay appeared in infancy. This review shows that the majority of mosicism forms affects fetal tissue, which, in turns, is associated with a high risk of fetal malformations. Furthermore, in 70% of cases fetal defects were so severe that termination of pregnancy was performed. In addition, we observed that 20% of neonates were born alive and well and only one infant presented mild defects. Mosaicism confined to the placenta appears to be associated with a better prognosis. According to our review, the partial involvement of the placenta might result in fetal growth restriction in about one third of cases, probably because the genetic anomaly cannot ensure a regular intrauterine growth. Only one fetus presented a major congenital heart disease, whereas most of the babies (72%) with placental mosaicism were normal and the unique neonatal death was related to prematurity rather than placental insufficiency. The finding that fetal mosaicism is more severe than placental mosaicism is further confirmed by the fact that when both the two conditions are present, 80% of pregnancies are terminated due to severe malformations. Fetal karyotype by umbilical blood cord sampling or fetal biopsy should be performed to differentiate placental from fetal mosaicism, although this approach might not be efficacious in all cases, because the mosaic cell lines might not involve fetal hematopietic cells. In addition, umbilical cord sampling and fetal biopsy might be complicated with infection, miscarriage and premature rupture of membranes. Parents should be counselled that when prenatal diagnosis of mosaicism is made, a definitive diagnosis to exclude or ascertain fetal involvement is not actually feasible.

Biography

A Cristina Rossi has received her Medical Degree in 1998 and Residency in Obs/Gyn in 2003. She is an author and reviewer of peer-reviewed journals. Her interest is in prenatal diagnosis and twin pregnancy. She is a Consultant in Obs/Gyn at Ospedale della Murgia (Bari, Italy).

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