

Morphological findings in malformed fetuses with normal karyotype

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In our Department morphological findings on fetuses from therapeutic interruption of pregnancy or spontaneous abortion are performed since ten years in order to correlate the ultrasound and/or chromosomal diagnosis with a real presence of malformations. The fetopathologic examination generally agrees with the chromosomal diagnosis, while in several cases it is possible to find malformations also in presence of a normal karyotype (Gitz, 2011).

In our experience over the past 5 years we have found that 17 fetuses with a normal karyotype showed different heterogeneous ultrasound malformations. Only in 2 cases the fetuses died in uterus (17th and 22nd weeks of gestation), the other cases, aged between 14th and 23rd weeks of gestation, went from voluntary abortions.

In 7 cases the karyotype was defined by amniocentesis while in the remaining 10 was determined by fetal fibroblasts culture; in only 30% of the observed cases the couple had carried out a genetic evaluation.

External malformations were present in 16 fetuses, often related to the face (such as micrognathia, low-set of ears, flattened nasal bridge, cleft lip) or limb (short, curved, stubby) of spine (spina bifida) or genitalia (hypospadias). Malformations of internal organs were present in 10 cases, often affecting the cardiovascular system (complex heart defects and abnormal origin of the great vessels), and nervous system (meningocele, agenesis of the corpus callosum, ventricular dilatation and Arnold-Chiari malformation); less frequent were malformations of other systems (digestive, respiratory and urinary). There was a single case of situs viscerum inversus associated with complex cardiac malformations and atresia of the bucco-pharyngeal membrane.

These results indicate that the fetal morphological study is useful not only to confirm but often to supplement and complete the ultrasound data.

Moreover genetic evaluation, utilizing fetopathologic study, may have an important role in defining the diagnostic and clinical procedure, especially in relapses with malformed fetus and normal karyotype.

References

Gitz L, Khung-Savatovsky S, Viot G, et al. Fetopathologic examination for early termination of pregnancy: dogma or necessity? *Am J Obstet Gynecol* 2011;205:467.e1-9.

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