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Prenatal screening and diagnosis of aneuploidy in multiple pregnancies

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Prenatal screening for aneuploidy has changed significantly over the last 30 years, from being age-based to maternal serum and ultrasound based techniques. Multiple pregnancies present particular challenges with regards to screening as serum-based screening techniques are influenced by all feti while ultrasound-based techniques can be fetus specific. Tests currently available tend to not perform as well in multiple compared to singleton pregnancies. Considerations must be given to these variations when discussing and performing screening for aneuploidy in this situation. Prenatal invasive diagnosis techniques in multiple pregnancies bring their own challenges from a technical and counselling point of view, in particular with regards to sampling error, mapping and assignment of results and management of abnormal results. This review addresses these particular challenges and provides information to facilitate care.

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Introduction

Multiple pregnancies present specific challenges with regards to prenatal screening and diagnosis. Although some of the usual components of screening such as previous history and family history remain an important feature of the screening process for aneuploidy, the various pregnancy specific screening methods present particularities when it comes to multiple pregnancies. Most of the

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information available on the topic covers twin pregnancies whereas the literature is much sparser on higher order multiples. The following paper reviews the literature available to facilitate counselling and health care resources planning on this topic.

Background

Zygosity and chorionicity

Zygosity is determined by the number of fertilized ova that resulted in the multiple pregnancy. It is assumed that monozygotic multiples (originating from a single fertilized ovum) have the same genetic make-up and consequently, should be concordant for aneuploidy. For screening purposes, this assumption is applied, although rare exceptions to this rule exist [1], described in case reports of heterokaryotypic monozygotic twins. Feti in a dizygotic or polyzygotic multiple pregnancy will have as many different genetic make-ups as there are zygotes (or fertilized ova). Consequently, these feti may be concordant or more frequently discordant for aneuploidy. As zygosity cannot be assessed directly without invasive testing during pregnancy, some inferences based on ultrasound findings must be applied, through the assessment of the chorionicity.

Chorionicity refers to the type of placentation and specifically to the number of chorions or functional placentas. It can be assessed by ultrasound, most successfully in the first trimester when the accuracy is between 96–100% [2–4]. Monochorionic multiple pregnancies are essentially always monozygotic. Most dichorionic or polychorionic multiple pregnancies are dizygotic or polyzygotic, with as many zygotes as there are chorions; less than 10% of them [2–4] will arise from a single zygote that divided within the first 3 days post-fertilization. As the majority of polychorionic multiple pregnancies are polyzygotic, it is assumed that they have different genetic make-ups and are most likely discordant for aneuploidy.

These concepts are particularly important when assessing the risks through screening and counselling on invasive procedures.

Prenatal screening in multiple pregnancies

Risk assessment based on maternal age, periconceptional factors and chorionicity

It is easy to conceive that the overall risk of aneuploidy is higher in multiple pregnancies compared to singletons. This is in part due to increased maternal age in multiple pregnancies (either spontaneous or conceived through fertility treatments) [5]. The maternal age to be used for counselling is the age of the egg at the time of retrieval for pregnancies arising from frozen embryo transfer or egg donation.

A monochorionic multiple pregnancy spontaneously conceived or conceived without the use of intracytoplasmic sperm injection (ICSI) carries a similar overall risk of aneuploidy compared to a spontaneously conceived singleton pregnancy for the same maternal age. Both twins will either be affected or not, with only rare exceptions as described above.

A polychorionic multiple pregnancy has an increased risk of aneuploidy as it is most likely polyzygotic and it is the number of zygotes that determines the level of risk for aneuploidy. Each fetus has, in theory, its own risk of aneuploidy, relatively independent of the risk for the other(s). Consequently, it has been suggested that the risks are additive. [6] For example, the risk of aneuploidy would be double that of a singleton in dichorionic multiple pregnancies, triple in trichorionic pregnancies and so on. This concept has been recently challenged [7] as the observed prevalence of Trisomy 21 in twin pregnancies is much less than the theoretical risk based on the above assumptions and as such, this simple rule must be applied with caution when assessing risk based on history alone. This is likely related to a variety of environmental and parental genetic factors that are not completely independent of each other and thus influenced all zygotes similarly rather than independently.

In view of the challenges described above and the trends towards using comprehensive screening rather than maternal age alone, as recommended by multiple national bodies such as Canada and Singapore [8,9], maternal age alone should be discouraged as a screening method for multiple pregnancies, unless no other methods are available. In that case, chorionicity should be considered, and the

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