Fetoscopy, fetal-tissue sampling and the ESHRE guidelines on prenatal diagnosis

Dear Sir,

In the otherwise laudatory document, ‘Investigation and treatment of infertile couples: ESHRE guidelines for good clinical and laboratory practice’ (ESHRE, 1995), the section on prenatal diagnosis of genetic disorders covers the topic of fetoscopy with a mere two sentences: ‘Fetoscopy may be indicated in selected cases of suspicious fetal abnormalities. It has never gained widespread use due to its high complication rate (Quintero et al., 1993)’. Although prenatal diagnosis may not have been the primary focus of the document, it is with mixed emotions that those of us involved in fetoscopy see the procedure dismissed so summarily. Actually, reasons for the less frequent utilization of fetoscopy are far more complex than suggested. In particular, one should distinguish between fetoscopy per se, a procedure that is indeed obsolete, and ultrasound-directed fetal-tissue sampling, a procedure spawned from fetoscopy and which still has a limited if ephemeral place in our prenatal diagnostic armoury.

The term ‘fetoscopy’ was first used by Scrimgeour (1976) to describe the technique of intrauterine visualization using endoscopic instruments. The procedure was initially conceived as a means of direct fetal visualization for such external fetal malformations as spina bifida, polydactyly, or skeletal dysplasias. A relatively large (25 mm diameter) trochar was needed to allow a fiberoptic endoscope to be introduced into the amniotic cavity for fetal visualization. Fetal blood sampling was possible through a side channel and, in the early 1980s, this was the only method of diagnosing fetal haemoglobinopathies. The fetoscope was soon adapted for fetal skin biopsy under direct visualization, permitting the prenatal diagnosis of genodermatoses, e.g. epidermolysis bullosa or the congenital ichthyoses (Elias et al., 1980). At that time, neither group of disorders was detectable by amniotic fluid or fetal blood analysis because the relevant genes were not expressed in those tissues. By the early 1990s, we and others showed that fetal skin biopsy could, like fetal blood sampling (cordocentesis), be performed under ultrasonographic visualization without the necessity for uterine puncture with the larger fetoscope (Elias et al., 1993). With concurrent ultrasound, only a 17 mm (14 gauge) catheter is required for introducing the biopsy forceps into the amniotic cavity. With ultrasound-directed fetal skin biopsy, our recent experience in diagnosing genodermatoses has been quite salutary (Elias et al., 1994). The same fundamental technique has been used by Evans et al. (1991) for fetal muscle biopsy in the rare cases in which DNA polymorphisms are not informative for detecting Duchenne muscular dystrophy.

Whether ultrasound-directed fetal skin biopsy has a lower procedure-related loss rate than fetoscopy is uncertain, but analogy with the relative safety of cordocentesis seems appropriate. However, in general, fetal tissue sampling is increasingly being abandoned because DNA-based diagnosis can make use of the safest possible invasive methods, often amniocentesis or chorionic villus sampling in experienced hands. Obsolescence of highly invasive procedures will doubtless continue. As one example of changing management, fetal Rh(D) status can now be made by DNA testing on amniotic fluid cells or even maternal blood (Bennett et al., 1993; Lo et al., 1993); thus, cordocentesis is not always necessary. For fetal skin sampling, the only remaining indications involve the decreasing number of genodermatoses in which a DNA-based diagnosis is still not possible.

Nostalgia for fetoscopy or any other invasive prenatal diagnostic procedure must not dissuade us from embracing newer, safer, and more accurate methods. Thus, we suspect the ESHRE working group would agree that the fetoscope should be viewed as a museum piece, which we can admire for having served our patients well in times past. However, before sealing the time capsule we should recall the fetoscope’s histological role and recognize it as being the progenitor of ultrasound-directed tissue sampling (skin, muscle, blood) and the progenitor of endoscopic fetal surgery, e.g. the endoscopically-assisted transuterine repair for fetal diaphragmatic hernia, fetal urinary tract obstruction, and twin–twin anastomoses which are now being attempted. Of course, these techniques, now in the next time capsule of prenatal diagnostic techniques, will also find themselves being replaced by ever-more sophisticated ultrasound, analysis of fetal cells recovered from maternal blood (Simpson and Elias, 1995) and DNA-based analysis of the most readily-available fetal nuclei.

Naturally all could be followed by gene therapy.

References


Letters to the Editor

Joe Leigh Simpson and Sherman Elias
Department of Obstetrics and Gynecology
Baylor College of Medicine
One Baylor Plaza
Houston, Texas 77030-3498, USA