

Mini-Symposium on Genetic Diagnosis

Preface

Hidekazu SAITO

Chief, Division of Reproductive Medicine (Sterility)
Department of Perinatal Medicine and Maternal Care
National Center for Child Health and Development
E-mail: saitou-hi@ncchd.go.jp

The incidence of major chromosomal abnormalities in newborns in Japan is approximately 0.7 percent. Prenatal genetic diagnosis (PND) has come to play an important role in detecting abnormalities before birth. Presently, there are three major procedures for the genetic status assessment of fetuses or embryos: amniocentesis, chorionic villus sampling (CVS) and preimplantation genetic diagnosis (PGD).

Four PND specialists were invited to this mini-symposium, and they will express their unrestrained opinions about the PND.

Dr. Sasabe will discuss the issue of PGD. He will talk about the advantages of PGD over classical PND methods, such as CVS or amniocentesis, and what genetic information can be obtained before implantation. He will also discuss diagnostic strategies, pitfalls and possible future developments of PGD in Japan.

Dr. Hashiba will also talk about PGD but focusing on the diagnosis of Duchenne muscular dystrophy (DMD). According to Dr Hashiba, there are three approaches for the PGD of DMD. The first approach is gender

determination of embryos by polymerase chain reaction (PCR) or the fluorescence in situ hybridization (FISH)-based method. The second is diagnosis of specific gene mutation by PCR with primers constructed to amplify the deletion exons. And the last is linkage analysis by the use of a linked marker.

Dr. Uehara will discuss CVS and its usage for prenatal diagnosis of the fetal karyotype through cytogenetic analysis, and of Mendelian inherited diseases through molecular or biochemical analysis. He will not only discuss the procedure but also the risks, the accuracy of the genetic diagnosis, the present utilization and future development of CVS in Japan.

Additionally, Dr. Sago will talk about amniocentesis, as it still is the most common invasive prenatal procedure for the detection of fetal chromosomal abnormalities. He will discuss the procedure, safety, indications of amniocentesis, the current usage and further improvement of amniocentesis in Japan.

I hope this mini-symposium will benefit all the members of our society in the present and future medical practices and in our basic researches.