CHAPTER I

INTRODUCTION

Amniocentesis is the most commonly performed invasive test for prenatal diagnosis of genetic disease, especially for chromosome abnormality(1,2).The standard amniocentesis is performed in the mid trimester between 16-18 weeks of gestation and the period from sampling to karyotyping is about 2-3 weeks. By that time the pregnancy is already evident, the mother feels the baby moving,and considerable "attachment" to the fetus already exists. The implications of an abnormal result at this gestational stage are emotional damage tc the parents and an increased maternal risk from pregnancy interruption at mid trimester. Early prenatal diagnosis offers the advantage of a safer termination of pregnancy with reduced social and psychological trauma. The period of waiting for the second trimester amniocentesis for prenatal diagnosis can cause the anxiety to the patient and the family. Stimulated by the pressure from the patient, first trimester prenatal diagnosis has been investigated including the chorionic villus sampling and early amniocentesis. However the first trimester prenatal diagnosis by chorionic villus sampling (CVS) is complicated by higher procedure related fetal loss

rate, more frequent maternal cell contamination, and placental mosaicism(3), and also increased risk of severe limb defects in the neonate(4). As an alternative to CVS, early amniocentesis at 11-14 weeks of gestation will be the procedure of choice for prenatal genetic diagnosis(5). Another advantage of early amniocentesis compared with CVS is the ability to interpret the amniotic fluid Alpha-Fetoprotein and Acetylcholinesterase for diagnosis of fetal abnormality(6,7).

The mean volume of amniotic fluid in early gestation for 8-13 weeks of pregnancy was about 15-90 cc(8). The effect of removal on excessive amount of amniotic fluid at this early stage may cause pregnancy loss and fetal deformity . But limiting the volume of the amniotic fluid specimen can cause low cell count and consequently cause culture failure and long duration of culture time for chromosome analysis(9,10).

This study deals with a new technique namely amnifiltration,used at 11-14 weeks of gestation.During aspirated the amniotic fluid is recirculated back to the amniotic cavity through a filter device,this will increase the cell yield and decrease the amount of fluid removed.This technique therefore is expected to improve the quality of the chromosome study especially the duration of cell culture

and the success rate of the result. The adverse effect to the pregnancy outcome and fetal complication related to excess removal of the amniotic fluid in the early stage of pregnancy should be decreased to the background rate.

The purpose of this study is to evaluate the quality of the cytogenetic result and pregnancy outcome in two groups of patients for early amniocentesis,those assessed with and without the filter technique.