A number of studies have looked at parameter as PAPP-A, free β- hCG, nicked β, urinary gonadotropin protein, SP 1, dimeric inhibin and ultrasound. This has resulted in a state of condition about the most likely best combination of parameters. By specifiying the demographic of the patient's age, ethnic background, maternal age a particular cocktail of parameters may be run
And finally, Chorion Villus Sampling, as a known invasive technic, may be used for the detection of chromosomal abnormalities in high risk groups isolated by biochemical analytes and ultrasound examination.

L140 *(Precongress Course)*

SONOGRAPHIC SCREENING FOR FETAL ANEUPLOIDIES

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First-trimester screening of fetal chromosomal abnormalities by chorionic villus sampling (CVS) was introduced at Szeged in 1982. The indication of the procedure was mainly the advanced maternal age (≥35 years) (85%). However, only a small proportion of children (7 % in Hungary, 17% in Finnland, 12.9 % in USA) were born to women age 35 years or older (Ventura, 2000). Following the maternal age indication as a guideline to screen aneuploidies, we were able to identify only a quarter of Down syndrome pregnancies, even if all women older than 35 years requested invasive procedures (CVS, amniocentesis). Consequently, advanced maternal age was not too good selection criterion for efficient prenatal screening of fetal chromosomal (and other) abnormalities due to the well-known controversy, that younger women have the majority of pregnancies, and younger women give birth to the majority of children with Down syndrome.
This controversy represented a great need for offering "some prenatal screening/diagnostic measures" to the younger (<35 yrs) pregnant population and we decided to develop a method for "in utero finding" defected fetuses irrespective of maternal age. We kept an eye on two criteria: the method should be unrisky and should effectively select pregnancies with fetuses of normal and abnormal karyotype.
The idea came from practice, namely: if a pediatrician could suspect Down syndrome by looking at the affected neonate on the base of trisomic features caused by "extra" chromosome, a sonographer should do the same by looking at the first trimester embryo. Further speculation was that the "extra" chromosomal material express more pronounced features in the early pregnancy. So we hypothesized that trisomic features predictive for trisomy 21 could be ultrasonically recognized as early as the first trimester.
The development of high-resolution ultrasound technique in the mid-80ths gave an outstanding opportunity to approach the intrauterine first-trimester embryo for detailed examination (sonoembryology). Therefore, from 1986 we examined each pregnancy with trisomy 21 fetuses, for finding some sonographic "attitude" that can differentiate aneuploid fetuses from ones with normal karyotype. Reexamining the trisomic fetuses with ultrasound an increased fluid accumulation in the fetal occipital and neck region had been found irrespective of the maternal age in a significant proportion of the fetuses with abnormal karyotype. The increased nuchal edema, which we called first-trimester simple hygroma (FTSH), enlighted the possibility of a prospectice screening in the general population .

L141 *(Precongress Course)*

INCREASED NT WITH NORMAL KARYOTYPE

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Introduction: Increased nuchal translucency marks not only the pregnancies complicated with chromosomal anomalies, but may also be found in fetuses with normal karyotypes. These pregnancies with increased NT and euploid karyotype may apparently be normal or candidate for certain structural or single gene defects.
Nuchal edema (NT) may progress or undergo spontaneous resolution during the late first and early second trimester both in euploid and aneuploid pregnancies. According to recent sonographic observati-

ons there is an association between increased NT and second trimester cystic hygroma, NT being the predecessor of the latter. This phenomenon provides basis for comparison of the natural history of simple hygroma in the first trimester and cystic hygroma in the second trimester of pregnancy.

Pregnancies with increased NT and normal karyotype have a high chance (more than 80%) of spontaneous resolution of the hygroma. Prenatal and neonatal progress has been reported uneventful in 89% of the cases, indicating a very good prognosis. However, cardiac and other structural malfrmations may occur in at least 20 per cent of the surviving embryos and more than 40 genetic syndromes have been reported up to now.

Second trimester fetal cystic hygroma, regardless of the underlying cause, carries a very poor prognosis and is correlated with unfavourable perinatal outcome. By contrast, increased NT may represent a similarly high risk of aneuploidy and an overall better prognosis in euploid cases. The improved prognosis for euploid fetuses with posterior hygroma detected in the first trimester may be related to the very early spontaneous resolution, since early resolution prevents irreversibile alterations.