PRENATAL DIAGNOSIS OF β-THALASSEMIA IN EGYPT

Elgawhary S*, Sahar M.Y. Elbaradie**, Wael Mohammed Rashad**, Maha Mosaad**, Abdalla M.A.H. **, Ezzat G* and Elbeshlawy A***
*Clinical pathology department, **Gynecology and Obstetric department and ***Pediatric department Cairo University, Egypt.

Abstract:
Objective: The clinical severity of thalassemia major and sickle cell syndromes make them priority genetic diseases for prevention programs through prenatal diagnosis for carrier couples. Incorporation of automated DNA sequencing that enable the characterization of mutations not detected by other mutation specific detection procedures was a prime concern of this work.
Study design: Automated DNA sequencing was offered on fetal tissues in ٣٠ pregnancies over the year ٢٠٠٥. The pregnancies were at high-risk for homozygosity or compound heterozygosity for β–thalassemia based on mutation analysis of both parents before prenatal diagnosis. Both parents were β - thalassemia-trait. Fetal samples were collected by chorionic villus sampling (CVS) in the first trimester and by amniocentesis in the second trimester. The point mutations were characterized by PCR (ARMS). The absence of the expected fragment with all the mutant ARMS primers insinuated an uncharacterized that was further subjected to direct automated fluorescent DNA sequencing in an attempt to know if the fetus is affected by parent's mutations.
Results: The mean gestation when carrying out the invasive procedure was ١٤ (١٠–١٨) weeks. All mothers had a previous affected pregnancy, and ١٣ had two or more previous affected pregnancies. Pregnancies were ٨ carrier fetuses (trait) and ٢٢ affected fetuses in which two were homozygous and ٢٠ double heterozygous. Parents in fourteen cases of the affected fetuses preferred to continue pregnancy and babies were born as diagnosed. The other ٨ decided termination and DNA of the abortuses proved to be as previously diagnosed by DNA sequencing.
Conclusion: the use of PCR amplification and direct sequencing have permitted the accurate characterization for unidentified alleles and successfully solved ١٠٠٪ of the examined samples.