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SAISIRI SITHONGDEE : PRENATAL DIAGNOSIS OF HOMOZYGOUS α–THALASSEMIA 1 BY USING FETAL NUCLEATED RED BLOOD CELLS IN MATERNAL CIRCULATION. THESIS ADVISORS: PATCHAREE WICHYANUWAT, M.D., Ph.D., PRANEE FUCHAROEN, Ph.D., SUTHAT FUCHAROEN, M.D., 97 p. ISBN974-663-702-9

This study examines the use of fetal Nucleated Red Blood Cells (NRBCs) in the diagnosis of homozygous α-thalassemia 1. This is the most common genetic disorder in Thailand. The 4 major thalassemic diseases are homozygous α-thalassemia 1, homozygous β-thalassemia, β-thalassemia in association with Hb E and Hb H diseases. Homozygous α-thalassemia 1 is the most severe form of thalassemia. Pregnant women with an affected child may counter certain complications such as toxemia of pregnancy. Prenatal diagnosis (PND) with selective abortion of the affected fetus is necessary. The procedures to obtain fetal cells for PND include fetal blood sampling by cordocentesis and chorionic villus sampling (CVS). These invasive procedures may create complications in both the fetus and pregnancy. Recent studies suggest analysis of NRBCs in the maternal circulation will prevent these complications. PND of homozygous α-thalassemia 1 using the fetal NRBCs is based on the immunological demonstration of the absence of normal α-globin chain in the fetal NRBCs with hydrops fetalis syndrome. The fetal NRBCs were isolated by density gradient centrifugation and enriched by immunomagnetic separation. The fetal NRBCs were further analyzed with fluorescent anti α-globin antibody.

Eleven high-risk pregnancies for Hb Bart's hydrops fetalis were studied using the fetal NRBCs in maternal blood. The blood was taken prior to CVS at 10-26 weeks of gestation. Eight out of eleven cases were non-hydrops fetuses, 3 showed the heterozygous α-thalassemia 1, 2 were Hb CS trait, 1 was heterozygous α-thalassemia 1 or Hb H disease and 2 were normal fetuses. Three cases were diagnosed to be Hb Bart's hydrops fetuses because no α-globin chain was detected in fetal NRBCs. All the results were confirmed with DNA analysis of chorionic villus. These results suggest that the immunofluorescent detection of fetal NRBCs enriched from maternal blood could be an accurate and sensitive method for screening of homozygous α-thalassemia 1. This procedure provides a noninvasive approach for PND of homozygous α-thalassemia 1.