



Sixteen-Year Experience of Prenatal Diagnosis of Thalassemia at a mainland Chinese hospital

Yin AH, Geng J, Du L, Qin DQ, Liang JQ, Wang YN
Guangdong Women and Children Hospital, Guangzhou, Guangdong, China

Objective

Thalassemia is one of the most prevalent hereditary diseases in south China. The gene frequency is about 16.8% in our local area, and it was considered a primary cause of the major birth defects in Guangdong province. Prenatal diagnosis plays an important role in preventing birth defect and improving birth quality. Through continuous efforts from the past sixteen years, we have established a hospital-based prenatal screening and prenatal diagnosis program successfully. Here we describe the experience of prenatal diagnosis of thalassemia at a Prenatal Diagnostic Centre in a mainland Chinese hospital.

Methods

From 1999 to 2015, we performed a total of 9052 prenatal diagnosis in pregnant women at risk for thalassemias. 23 mutations were tested routinely, DNA sequencing was performed when no mutations were detected.

Results

In total, 2232 tests have been performed by chorionic villus sampling (CVS), 6143 by amniocentesis and 674 by percutaneous umbilical blood sampling/fetal blood sampling. 6583 couples were α -thal alone, 2426 couples were β -thal alone, 30 couples were carriers of both α - and β -thalassemia, 11 couples who tested α -thal or β -thal or $\alpha\beta$ -thal in one partner and heterozygote of hemoglobinopathy in the other were found. 2185 fetuses were found to be normal, 4556 fetuses were carriers of the trait, 898 were detected Hb H disease, and 1413 fetuses were Hb Bart's hydrops fetalis or β -thalassemia major.

Conclusion

Prenatal diagnosis of thalassemia plays an important role in prevention and control the birth of fetus with medium and severe thalassemia. A number of Hb H disease and severe thalassemia have been prevented during the past 16 years of operation.