17th World Congress in Fetal Medicine

Development of a non-invasive prenatal screening for fetal aneuploidies using next-generation sequencing in Vietnam

Nguyen VT, Vo Thanh B, Trinh NTH, Nguyen MT, Nguyen HN, Nguyen TQT, Hoang TDT, Truong DK, Do TTT, Giang H, Phan MD, Nguyen HN

Hung Vuong Hospital, Ho Chi Minh City, Viet Nam

Objective

Non-invasive prenatal testing (NIPT) for fetal aneuploidies has been widely adopted in developed countries. This method has shown high sensitivity and specificity, helping to reduce the number of invasive procedures and increase health care quality for pregnant women. In Vietnam, collected blood samples have been sent overseas to perform the test with high cost and lengthy turnaround time. This study aims to develop a complete non-invasive prenatal testing protocol in Vietnam for fetal aneuploidies using maternal blood.

Methods

An application study. Blood from high-risk pregnant women with singleton pregnancies in the second trimester was collected for this test before proceeding to amniocentesis at Hung Vuong Hospital. The complete protocol was performed: cell-free DNA extraction from blood, preparation of sequencing libraries, sequencing using Illumina massively parallel sequencing system and data analysis using an improved size-based algorithm called triSure.

Results

Our protocol was successfully established and applied to 141 samples from high-risk pregnant women. The results were compared with the results of karyotyping at Hung Vuong Hospital. Among 61 samples confirmed to have aneuploidy by karyotyping, 43 of which were trisomy 21, 15 were trisomy 18 and 3 were trisomy 13, our protocol could identify all of them, showing a concordance of 100% in result between our NIPT protocol and karyotyping.

Conclusion

This study has successfully developed a complete protocol for non-invasive prenatal screening for fetal aneuploidies. The application of this NIPT protocol in Vietnam could reduce the cost and turnaround time, providing a high-risk screening option, in order to decrease the need for an invasive diagnostic test.