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Fetal RHD genotyping in maternal plasma: 4 Years of experience

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Background: Fetal RHD genotyping in maternal plasma has been carried out for 4 years in the laboratory.

Aims: The analysis involves three of the 10 exons of this gene, exons 4, 5 and 10 amplified by real-time PCR and can be performed from 10 weeks of gestation. The criteria for technical and biological validation have been defined: Positive results in case of amplification of the three exons, negative results in case of no amplification (that must be controlled on a new sample taken 15 days later), undetermined in other cases. The expression of the results is standardized with well-defined biological comments. A tracking sheet of pregnancy is attached to the report to obtain from the maternity the results of RH1 phenotype at birth (obtained by immunological method). It is very important to verify the absence of false negatives by this method.

Results: Since 2010, 64 analyzes were performed: 38 results were positive, 14 negative and 12 undetermined usually corresponding to a maternal or in several cases fetal RHD gene variant.

Conclusion: The experience of almost two years shows that this method with procedures and very precise and rigorous criteria of validation is reliable. Situations of maternal variants are the most difficult situations that should lead to careful interpretation of the results. The laboratory participates to the elaboration of a quality control.

Biography

I am an M.D/Ph.D in Biology. I work currently in a laboratory of a Health Clinical Center, CHU of Lyon. I specialize in immuno-haematology as well as in NIPT: fetal RHD genotyping on maternal plasma. I was invited to present this at several international conferences (ESHG 2012) and I am the author of a recently published article (Non-invasive fetal RHD genotyping: Validation of the method with 200 patients, TCB, 2014).

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