

Non-invasive prenatal screening for diagnostics for chromosomal abnormalities, and it's relation to forensic practice

Putzová Martina¹, Paulasová Petra², Šprdlíková Lucie¹, Milan Macek², Michal Michal¹

¹ Bioptická laborator, Pilsen, Czech Republic

² FN in Motol, Institute of Biology and Medical Genetics

Background: Non-invasive prenatal diagnosis is currently part of routine practice and management in the early stages of pregnancy. The Panorama[®] test (Natera, US), is the only available test based on targeted sequencing of selected areas and their subsequent analysis. Thanks to this technology, the Panorama[®] is unique in several ways: it is the only one that can reliably distinguish free fetal DNA from maternal DNA. In the presentation, the author deals with an unusual case, which was initially delivered to the laboratory as a request for NIPT analysis in surrogacy, but the Panorama[®] test, disproved surrogacy and started unraveling of very problematic case with a series of paternity analyses.

Methods: Panorama[®] is the only NIPT test whose entire evaluation is based on the analysis of SNPs. This approach is the only one that allows detecting the origin of the fragments and thus distinguishing the maternal from the fetal fraction. Since 2020 the examination is available also for patients with donated oocytes and twin pregnancies.

Results and Conclusion: Author deals with an interesting case, which was referred to the laboratory as an NIPT of surrogate mother, but the SNP evaluation was not compatible with surrogacy. Surrogacy is in Czech Republic legal, must be indicated for medical reasons. There is no control mechanism/ register in Czech Republic of woman offering surrogacy. From the mapped case is obvious, that in lack of control, some people make a good business out of surrogacy. They utilize the fact, that affected individuals or couples, usually do not take legal actions, because of their emotional vulnerability. In this case the employed NIPT based on SNP testing (Panorama[®], Natera, US) was, because of its methodology, which enables the deep insight into each tested pregnancy case, the first one to reveal the discrepancy in the case history of the patient.