Modern High Throughput Approaches are not meant to Replace ‘Old Fashioned’ but Robust Techniques

Thomas Liehr*, Kristin Mrasek, Elisabeth Klein and Anja Weise
Institute of Human Genetics, Jena University Hospital, Friedrich Schiller University, Jena, Germany

Genetics and Genomes: A Brief Discussion

Presently, there are major discussions in the field of genetics and genomes if some approaches may be outdated and to be replaced by more recent and modern ones. Especially if it is on human genetics, one easily finds papers claiming that e.g., banding or molecular cytogenetics could or should best be replaced by array comparative genomic hybridization (aCGH) and next generation sequencing (NGS) technologies [1-5]. Often there is also the claim that aCGH is cheaper and NGS more reliable than the ‘old-fashioned’ approaches, even though both allegations already showed to be not true [6,7].

Besides, one should remember that in most countries around the world it will be practically impossible to adapt aCGH and NGS during the next decades due to limited financial resources. Furthermore, it is questionable if e.g., a clinically suspected Down syndrome should really be characterized by a high throughput approach instead of single cell oriented techniques like cytogenetics. Apart from cost efficiency aspects, for correct genetic counselling a translocation trisomy 21 must always be distinguished from free trisomy 21 - thus chromosome analyses cannot be skipped in such cases, as neither aCGH nor NGS can separate them.

Just to pinpoint the misleadingness of some of the recent papers, exclusively glorifying new, and condemning old approaches we compared our own data on detection rates in a specific application achieved with ‘standard methods’ to data of a recent ‘glorifying’ paper [1]. The latter [1] claimed, that in miscarriage-samples from women who had an early spontaneous abortion, the application of SNP-aCGH and NGS would lead to most comprehensive results with detection rates of ~50%. In our laboratory, we do a combination of banding cytogenetics and microsatellite analyses of maternal blood and DNA isolated from abortion-tissues to answer this question. Surprisingly, we found comparable detection rates of ~48% as when using much more expensive and not everywhere available high throughput approaches.

In detail: By banding cytogenetics we studied overall 237 tissues derived from miscarriages. 50 of them did not show any growth (18.3%), 28 revealed normal male (11.8%), 74 normal female (31.2%) and the remainder 85 abnormal karyotypes (38.7%). In a second step only those cases without a result explaining the miscarriage (i) and those with normal female karyotype (ii) were studied by microsatellite analyses. Group (i) was studied to detect triploidy, trisomies or monosomies and exclusively brand-new approaches. Especially, when it is not about research but about diagnostics this is an important point. Some conservativeness in staying with and remembering the advantages of the well-established standard approaches supports usage of available resources thoughtfully.

References


*Corresponding author: Thomas Liehr, Institute of Human Genetics, Jena University Hospital, Friedrich Schiller University, Postfach, D-07740 Jena, Germany, Tel: +493641935533; Fax: +493641935582; E-mail: Thomas.Liehr@med.uni-jena.de

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