Molecular Blood Group Diagnostics

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This and the next issue of TRANSFUSION MEDICINE AND HEMOTHERAPY draw special attention to ‘Molecular Blood Group Diagnostics’ and ‘Molecular Diagnostics in the Whole Genome Age’, respectively.

Scientific reports related on molecular blood group diagnostics by scientists from German speaking countries are significant measured by impact and numbers. However, this special issue of TRANSFUSION MEDICINE AND HEMOTHERAPY will refrain from focusing on ‘historical’ findings and rather address timely topics: This issue reviews on different methods for medium- to high-throughput DNA typing methods, precises currently debated routine applications for blood group genotyping, and puts the question: ‘Will genotyping replace serology in future routine blood grouping?’

Looking back for the last two decades, DNA typing in molecular blood group diagnostics widely took benefit from technologies established and tested for HLA typing in the first place. Still, peering over the ‘HLA shoulder’ results in interesting technology transfers as illustrated in this issue for the Luminex® methodology by Drago et al. [1]. On the other hand, there are different specifications in blood group genotyping with respect to sample numbers and gene polymorphism as compared to HLA. This might explain, why chip-based DNA typing is adopted sincerely by blood group specialists, as being detailed in two contributions by Avent et al. [2] and Reid [3] in the current issue. The key aspect of methods for medium- to high-throughput DNA typing methods is rounded up by a review report on MALDI-TOF mass spectrometry and its fascinating potential for blood group DNA typing by Garritsen et al. [4].

Today, employment of molecular blood group diagnostics for the genetically exact definition of blood group variants (e.g. *RHD* and *ABO*), clarification of original genotypes in poly-transfused sample material, or antibody-masked erythrocytes are generally accepted. However, there are other attractive applications of this methodology, e.g. in testing a fetus’ *RHD* type

from maternal serum. Referring to this, Legler et al. [5] present a timely review of current European efforts. Cost-effective screening for donors with rare antigen profiles represents another up-to-date topic and is picked out twice as a central one in this issue by Jungbauer [6] and Wagner [7]. Since nobody can do without quality control nowadays, an example of how molecular blood group diagnostics might influence serological validation processes is presented by Gassner et al. [8]. No matter which method is being used and blood group polymorphisms are being analysed, there will always be a certain percentage of samples remaining inconclusive when combining serological and molecular biological results. In most cases, these results will present new blood group alleles, and there is a vital interest for resolving these cases by means of DNA sequence-based typing as being detailed by Seltsam and Doescher [9].

Five expert opinions on the question ‘Will genotyping replace serology routine blood grouping in the future?’ represent a third key aspect of this issue [10–14]. Experts are being challenged by giving editorial sub-queries such as: ‘Should there be genotyping for ‘all’ (currently available) blood groups of all donors? Could there be additional testing for genetic markers encoding ‘soft’ genetic diseases such as e.g. haemochromatosis, hereby extending responsibility range of blood typing centres in the context of public health? Could there be a blood group serologists’ approval for ‘in silico’ cross-matching, given all available blood groups of each transfusion event were genetically determined?’

Browsing the content of this issue, most of the technical applications which are being presented are developed and tested in highly specialised laboratories only. Some of them will always remain there. Others will might be picked up by the industry and made accessible to the end users. In any case, skilled experts in molecular biology and intense cross-talk to medical specialists are two irreplaceable prerequisites for an innovative application of modern molecular blood group diagnostics.

References

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