100 Years after von Dungern & Hirschfeld: Kinship Investigation from Blood Groups to SNPs

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This issue of TRANSFUSION MEDICINE AND HEMOTHERAPY is dedicated to the pioneers in immunohematology and forensic genetics and in particular to the Polish physician and immunologist Ludwig Hirschfeld (fig. 1). Born August 5, 1884 in Warsaw, he died March 7, 1954 in Wrocław. From 1907 to 1911 Hirschfeld worked at the Heidelberg Institute for Experimental Cancer Research together with Emil von Dungern. In 1910 they published their fundamental work on the inheritance of ABO blood groups [1] that is recognized as a milestone in human genetics and the onset of forensic genetics.

In the 20th century the progress of forensic genetics was mostly affected by the introduction of the inheritance of the ABO and further blood group systems and later by genetic polymorphisms of serum proteins, leukocyte antigens (HLA), and erythrocyte enzymes. The era of anonymous DNA markers, i.e. the tandem repetitive DNA regions (minisatellites), was heralded in 1985 when Sir Alec Jeffreys and colleagues [2] presented the DNA fingerprinting technique. Enormous progress was then achieved when PCR-based analysis of short tandem repeats (STRs) was introduced in the early 1990s. The history of forensic genetics and the use of the different genetic marker systems in kinship analysis are reviewed by Geserick and Wirth [3] in this issue TRANSFUSION MEDICINE AND HEMOTHERAPY.

Since DNA sequence analysis of different genomes was intensified, researchers became aware of diallelic markers that represent the most abundant genetic variation evenly spread in the genome. These single nucleotide polymorphisms (SNPs) and short insertion-deletion polymorphisms (indels) are the basis of current developments in forensic genetics that are summarized in the article by Schneider [4]. The diallelic marker systems and their use in trace and kinship analysis is also the topic of the original research articles in this issue. One option could be the typing of non-anonymous SNPs with a clear genotype-phenotype correlation such as SNPs in the

Fig. 1. Ludwig Hirschfeld (Ludwik Hirschfeld; from <http://zlotemysli.w.interia.pl/sentencje/h/hirschfeld.html>)

ABO gene representing the molecular basis of the ABO blood groups [5]. A larger panel of anonymous autosomal SNPs, the 49 SNPforID SNPs, is already used in different laboratories. Experiences are presented by the groups of Schwark et al. [6] and Børsting et al. [7]. Furthermore, Phillips et al. [8] report about the application of both the SNPforID panel and high-density SNP arrays including about 1.8 million SNP and indel markers. The current progress of introducing diallelic markers to forensic genetics is amended by the work of Pena and Pena [9] who describe a powerful and low-cost method for typing of 40 indels with a widespread chromosomal location.

Over the past 100 years forensic genetics including kinship analysis has developed from phenotyping of blood groups and other blood cell markers to genotyping of anonymous DNA markers. This progress was mostly driven by the significantly

higher power of exclusion for the DNA markers, especially the highly polymorphic STRs. However, STR-based analysis is hampered by the relatively high mutation rate of the markers. This is one of the driving forces for the development and

introduction of DNA fingerprints based on the much more stable diallelic markers. All together, these achievements led to the current status of paternity testing with clear and convincing evidence due to highly informative genetic systems.

References

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