J. Clin. Chem. Clin. Biochem.
Vol. 25, 1987, p. 121

Editorial

Gene Diagnostics — A Challenge for Clinical Chemistry?

Gene technology has greatly improved the prenatal diagnosis of sickle-cell anaemia and other haemoglobinopathies in recent years. Using DNA-probes or oligonucleotides the analysis for mutations can be carried out after 8—10 weeks of pregnancy with chorionic villi samples or after 16 weeks of pregnancy with amniotic fluid. DNA recombinant technique is now spreading so fast that other common inherited disorders may be analysed in the near future. The fact that restriction fragment length polymorphisms (RFLPs) can be used as genetic markers has opened up a new dimension for the study of the human genome. This technology is becoming increasingly applicable to the detection and diagnosis of other important monogenic disorders — even where the biochemical defect is not yet understood. Diseases such as cystic fibrosis, *Duchenne*'s muscular dystrophy, *Huntington*'s chorea, steroid 21-hydroxylase deficiency, X-linked retinopathies, haemophilia A and B are now amenable for gene diagnostic study if a DNA sample is available from an affected or normal sib in the family. Genetic markers could also be used in the prediction of atherosclerosis, diabetes mellitus and even risk factors of otherwise undetectable multigenic diseases. Techniques that are currently being developed, such as non-radioactive detection of DNA sequences and pulse field electrophoresis for the separation of DNA molecules up to several million basepairs will play an increasing role in the future. The new aspects have been discussed at the 2. Annual Meeting of the Arbeitsgemeinschaft für Gen-Diagnostik e. V., the abstracts of which are part of this issue.

For the classical clinical chemist, gene techniques may merely mean the introduction of a new analytical technique like radioimmunoassay, isoelectric focusing or cell differentiation by surface markers. This view however, ignores the potential of gene analytical techniques. Besides opening the possibility of replacing hitherto measured products by direct gene analysis, these methods allow for qualitative results long before gene products are formed and the abnormality or disease is expressed.

For this reason the questions may be raised "what should be measured", "when should be measured" and "in what kind of population" leading to general ethical problems which cannot be solved by the gene analyst alone.

It is hoped that this and the forthcoming annual meetings of the "Arbeitsgemeinschaft für Gen-Diagnostik e. V." provide a glimpse of the potential molecular approach to disease and allow interdisciplinary discussions about the technical, medical, ethical and social aspects of the new technology.

A recent report of the Enquete-commission: "Chances and Risks of Gene Technology" (Drucksache 10/6775 of Deutscher Bundestag 6. 1. 1987) summarizes the possibilities and gives recommendations for limitations of gene analysis in prenatal and postnatal diagnosis, in the screening of employees and in insurance screening as well as in jurisdiction.

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