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特 別 講 演 Special Lecture

**Retrospects and Prospects of Research on Inborn Errors of Metabolism, as Exemplified
by Phenylketonuria**

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Since the basic work of Garrord in 1908 on four metabolic disorders more than 150 well defined inborn errors have been published. Most of them are inherited as autosomal recessive traits, and they produce a very broad spectrum of symptoms and organic lesions, involving the brain, liver, kidneys, bones, skin, eyes, intestinal tract *etc.* For an increasing number of these disorders therapeutic approaches have been developed. For some, effective therapy is now available, whilst for others, treatment is still on trial. A delayed diagnosis usually results in damage to various organs, especially to the brain, which is increasingly irreversible and often fatal. This stresses the importance of an early diagnosis, ideally by screening all newborn infants of a population by special screening programmes. Mention will also be made of diseases which are amenable to treatment but for which no screening procedure permitting a timely diagnosis is yet available, *e.g.* fructose intolerance, Wilson's disease and the adrenogenital syndrome. It will be a matter of great importance for future research to develop suitable screening techniques for as many treatable diseases as possible.

Means of *true* prevention of hereditary enzymopathies and proteinopathies are at present still of limited practical value though of great importance for the future. They have been thoroughly discussed by a World Health Organization scientific group on prevention, treatment and rehabilitation in genetic disorders, the report of which has been published in 1972. They include techniques such as prospective and retrospective genetic counselling, contraception, artificial insemination with donor semen, pregnancy termination after a prenatal diagnosis using amniotic fluid or cells, sterilisation, *etc.*

Of the many inborn errors of metabolism, phenylketonuria is the best example. For this reason I will in my lecture concentrate on this disease. In many hundred publications this disease has been studied most thoroughly. It has stimulated similar research and advances in other metabolic disorders with or without cerebral involvement, especially as regards early diagnosis by screening and therapy. So phenylketonuria, though most important in itself, has become a model for the more than hundred other inborn errors of metabolism.