

Screening and management of potentially treatable genetic metabolic disorders

Editor P. F. Benson

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Screening And Management Of Potentially Treatable Genetic Metabolic Disorders:

Screening and Management of Potentially Treatable Genetic Metabolic Disorders P.F. Benson, 1984-03-31 1 The success of early diagnosis and therefore of treatment of phenylketonuria inevitably suggests the possibility of early screening and treatment of other genetic metabolic abnormalities This volume contains a summary of papers delivered at a Workshop on Recent Advances in Screening and Management of Potentially Treatable Genetic Metabolic Disorders held under the auspices of the Commission of the European Communities in London U K on the 17th and 18th March 1983 to consider such possibilities The Workshop was not aimed at those disorders for which the principles of treatment and management have been soundly established or for which screening procedures are in general use The papers therefore do not form a comprehensive account of metabolic disorders The topics for discussion were selected mainly to highlight recent discoveries which might be exploited by concerted approaches between different centres especially when the immediate benefits were restricted to only a few centres Recent reports suggest that about 2% of infants with persistent hyperphenylalaninaemia do not respond to treatment by phenylalanine low diets and develop severe brain damage Infants with such Malignant Hyperphenylalaninaemia due to one of several genetic causes of tetrahydrobiopterin deficiency should be rapidly identified since there is evidence that appropriate treatment will prevent brain damage The Workshop considered how appropriate screening could be made generally available

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Communities,1984 **National Library of Medicine Current Catalog** National Library of Medicine (U.S.), **Antenatal Diagnosis of Fetal Abnormalities** James O. Drife,Dian Donnai,2012-12-06 In few areas of medicine is progress more spectacular than in the field of prenatal diagnosis New clinical techniques such as chorion villus sampling detailed ultrasound scanning and cordocentesis are being evaluated by obstetricians and refinement of biochemical testing is widening the scope of maternal serum screening In the laboratory dramatic advances in molecular biology are occurring families at risk of genetic disease can be investigated with gene probes and preimplantation diagnosis of the embryo is now becoming a reality These technical advances have important ethical and practical implications among which will be a further increase in public expectations of the standards required of antenatal services Clinicians will need a high degree of skill to inform healthy women about the options for screening normal pregnancies and to counsel high risk women about the benefits and limitations of prenatal diagnosis Obstetricians scientists and health service managers will face the difficult task of deciding how prenatal diagnosis can be made available to women in a caring and cost effective way Recognising the rapid progress in this field the Royal College of Obstetricians and Gynaecologists made prenatal diagnosis the subject of its 23rd Study Group An international panel of leading researchers whose expertise ranged from molecular biology to philosophy was invited to participate in a three day workshop with time for in depth discussion as well as the presentation of papers

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