reference


Much has been written about the decline of the adult necropsy. Apart from paediatricians, few had recognized that the paediatric necropsy is, in fact, a genetic investigation which, taken in conjunction with antenatal diagnosis, is in reality a contributor to preventive medicine. The authors summarized the more interesting results obtained by the study of cytogenetics, tissue culture, and enzyme chemistry in the field of paediatric pathology, and the application of the findings in antenatal diagnosis. Special consideration was given to two aspects. First, results were presented of routine chromosome analysis on two series of consecutive perinatal necropsies, revealing that in up to 10% of these, there had been a chromosome abnormality. Secondly, the importance of accurate diagnosis of metabolic disorders at the specific enzyme level was illustrated by the current antenatal diagnosis of certain lysosomal enzyme deficiency diseases, such as metachromatic leucodystrophy, Sandhoff's disease, and Tay Sachs disease.
A D Bain, G A Machin, G R Sutherland and J Butterworth

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