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Raising awareness and funding for brain tumour research

THE PMS2 GENE AND ITS ROLE

Dr. E. Sheridan, Leeds.

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Primitive neuroectodermal tumours (PNETs) are an uncommon but highly malignant type of childhood brain tumour. The term can be used to encompass Medulloblastomas (which usually arise in the cerebellum) as well as the rarer supratentorial PNET (arising in the cerebral hemispheres).

Researchers have recently discovered that in two families in which supratentorial PNET occurred in more than one child, the underlying cause is an inherited defect of a gene called PMS2. Other types of brain tumours also occurred less commonly in these families, including lymphomas.

The normal function of the PMS2 gene is to repair errors in the DNA sequence that arise during the course of growth and development. Our studies showed that genetic analysis of PMS2 is very difficult, and that previous work on PMS2 may have failed to detect important abnormalities in the gene, either for technical reasons, or because the wrong types of cancer were looked at.

The researchers on this project propose to (a) address the question of how frequently PNET results from alterations in PMS2, and (b) develop new methods of testing for abnormalities in this gene, and for cancer predisposition of the kind seen in the families they have encountered.