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Comments and Critique

Constitutional Aplastic Anaemia: A New Concerted Action for European Fanconi Anaemia Research (EUFAR)

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RARE MUTATIONS have often been at the start of new scientific developments. Human mutations that cause cancer-prone chromosomal instability disorders, such as Fanconi anaemia (FA), could be of extreme importance for understanding subcellular mechanisms, involved bone marrow function as well as in the avoidance of cancer. Patients suffering from FA develop a severe aplastic anaemia and are cancer-prone. FA cells are also exceedingly sensitive to bifunctional alkylating agents (such as cyclophosphamide, cisplatinum, mitomycin C). The syndrome was first described in 1927 by the Swiss paediatrician G. Fanconi. The recent identification of the first FA gene by the laboratory of Dr M. Buchwald (Hospital for Sick Children, Toronto, Canada) has generated a new surge of interest in this syndrome. The first FA gene is novel and the deduced amino acid sequence does not contain any motif that could reveal something of its function in the cell, so that major discoveries may be expected. EUFAR, supported by the Commission of the European Communities, has been established to streamline European developments in FA research.

EUFAR integrates ongoing FA research in Europe, including laboratories in France, U.K., Germany, Italy and the Netherlands. The project leader is Professor Eliane Gluckman, Hôpital Saint Louis, Paris.

The objectives of the concerted action are: (1) to determine the number of genes involved in FA, to clone these genes and to determine their genomic structure, regulation and function(s). (2) To determine the FA gene mutations found in European FA patients. (3) To correlate genotypes (specific FA gene affected; nature of the mutations found) with cellular characteristics, clinical phenotype and prognosis. (4) To seek application of the results for the benefit of FA families, i.e. carrier detection, prenatal diagnosis, clinical trials, improved bone marrow transplant procedures and development of gene therapy.

There are two centralised facilities to support the concerted action: (1) a registry of all FA patients diagnosed in the participating countries, (2) a cell repository for FA lymphoblast lines, as a source of reference material.

During the action period, EUFAR will organise workshops and issue a newsletter. For more information, please contact the project-leader (Prof. dr. E. Gluckman, Unité de Greffe de Moelle, Hôpital Saint Louis, 1 Avenue Claude Vellefaux, 75010 Paris Cedex 10, France; Tel: +33-1-42.49.96.44, Fax: +33-1-42.49.96.34) or the secretariat (Dr H. Joenje, Department of Human Genetics, Free University, Van der Boechorststraat 7, NL-1081 Amsterdam, The Netherlands; Tel: +31-20-54.82.764, Fax: +31-20-54.83.329).

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