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Assessment of quality of genetic counseling services by study of adverse events. R Harris & Hilary J Harris, University of Manchester, England

Quality assessment of genetic counseling is rare in the 31 European nations of the Concerted Action on Genetic Services (CAGSE*). Quality is here defined as the documented provision of genetic counseling in individual patient's records and adverse events are here defined as the occurrence of preventable genetic disorders when records suggest poor quality counseling has denied patients informed and unconstrained decisions. A lack of quality assessment is a matter of particular concern when counseling is provided by physicians who are not trained in medical genetics. In the UK a national audit** has been successfully completed of adverse events associated with easily recognisable high-risk situations. Non-geneticists provided genetic counseling because only a minority of such cases is referred to clinical geneticists. The adverse events included within a defined period all Down syndrome pregnancies in women of 38 or more, all live born infants with neural tube defect, cystic fibrosis in a second sibling, beta thalassaemia major pregnancies and late onset medullary carcinoma of the thyroid (multiple endocrine neoplasia, MEN2). The overall findings were that non-geneticist clinicians concentrate on the management of disease and may overlook the need for counseling and recording data which patients will later need for decisions about reproduction or disease prevention. Counseling, screening and prenatal diagnosis were sometimes impossible because of late booking in pregnancy or because of delayed diagnosis of an earlier affected child with CF. There are marked regional inequalities of access to genetic services particularly for minority ethnic groups with increased risks of thalassaemia. Ongoing audit of adverse genetic events, including all cases of early onset breast and bowel cancer, is being developed as part of a national clinical governance strategy to improve the understanding and use of genetic counseling services.

* Genetic Services in Europe. A comparative study of 31 countries by the Concerted Action on Genetic Services in Europe. Eur J Hum Genet 1997;5(suppl 2); 1-220.
** Harris R, Harris HJ, Lane B, Alberman, E, Williamson P, Dodge J, Modell B, and Wald N. How much genetic counseling do non-geneticists do? Overview of results of National Confidential Enquiry into Counseling for Genetic Disorders. J Roy College of Obstetricians and Gynaecologists (in press)

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Variable expressivity of Familial Medullary Thyroid Carcinoma (FMTC) due to a *RET* V804M (GTG→ATG) mutation in two families: Reluctance of gene carriers to accept prophylactic thyroidectomy. G.L. Feldman[1], I. Schuffenecker[2], G.M. Lenoir[2], A.W. Saxe[3], J.R. Roberson[1], N. Petrucelli[1] and C.E. Jackson[1]. [1]Henry Ford Hospital, Detroit, MI, [2]Hôpital Édouard Herriot, Lyon, France, [3]Sinai Hospital, Detroit, MI.

Multiple endocrine neoplasia type 2 (MEN 2) and familial medullary thyroid carcinoma (FMTC) are autosomal dominantly inherited cancer syndromes that predispose to the development of C-cell hyperplasia and MTC. MEN 2A and FMTC are caused by multiple mutations in exons 10, 11, 13, 14 and 15 of the *RET* proto-oncogene. Associations between phenotype and specific germline *RET* mutations in exons 10, 11 and 13 have been established. We have studied 2 large FMTC families with a mutation in codon 804 in exon 14 (GTG→ATG; Val804Met) in which some individuals over age 70 had no overt clinical evidence of medullary thyroid carcinoma despite carrying or transmitting this mutation. In addition, some affected individuals with metastatic disease have had a relatively benign course, suggesting that this mutation causes a less aggressive form of MTC. Because of the presence of active MTC, including metastasis, in younger individuals within these families, we have continued to advocate thyroid surgery on the basis of mutation analysis. However, the apparent absence of disease in older individuals caused some of the younger individuals also known to be gene carriers to be reluctant to accept the recommended prophylactic thyroidectomy. Thus, counseling issues remain difficult in families with FMTC for specific mutations, even in a genetic disease usually considered to have a high penetrance, often with expression at much younger ages. These families emphasize the importance of molecular genetic testing in individuals or families with a history of FMTC, since identifying the specific RET mutation significantly influences the decision-making process for the affected individuals.