

Letters to the Editor

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Minimum Guidelines for the Delivery of Clinical Genetics Services

To the Editor:

During this century numerous laws and programs have been introduced for the regulation and evaluation of physicians and the care they provide. Although much of this regulation and assessment has been done by private organizations and the states, the federal government has gotten involved primarily through Medicare patient evaluations by peer-review organizations and through control of reimbursements (Tokarski 1989). On the other hand, little has been done with regard to the evaluation of ambulatory care that physicians provide. A notable exception to this situation is the provincewide peer assessment of physician's office practices in Ontario, Canada (McAuley and Henderson 1984; Davis et al. 1990). Apparently, even less has been done regarding the quality of care provided by clinical geneticists, genetics counselors, and clinical genetics centers in general. In this letter, we present minimum guidelines, with the hope that they will be used to establish standards of care for clinical genetics services.

We have developed these guidelines as an ongoing effort of the Evaluation of Clinical Services Subcommittee, a committee within the Great Lakes Regional Genetic Group (GLaRGG), to provide assessment of the clinical genetics units in our six-state region. The GLaRGG region consists of Illinois, Indiana, Michigan, Minnesota, Ohio, and Wisconsin. These guidelines, known as the "Minimum Guidelines for the Delivery of Clinical Genetics Services," are now being used to evaluate centers within our region. Each center participates

in the evaluation process on a voluntary basis. The on-site evaluation is done by a volunteer peer reviewer from another state within the region.

We publish these guidelines in the hope that they will stimulate discussion on the need for evaluation of clinical genetics services and will serve as the basis for developing nationwide standards for such services. We welcome and encourage comments about these guidelines and about the wisdom of undertaking quality assurance of clinical genetics services.

The "clinical genetics unit," or simply the "unit," refers to whatever has been administratively designated as an entity that provides clinical genetics services; it can be a subsection of a section, a section or division within a department, a department within a school, or a private institution, office, or agency. It also should be noted that the subcommittee's guidelines, stated below, are general recommendations for the practice of clinical genetics, and a center's having met these guidelines does not necessarily assure a good quality of care or a positive patient/family experience or outcome.

The following guidelines must be met by any genetics unit providing clinical genetics services:

1. An American Board of Medical Genetics–certified or –qualified M.D., D.D.S., or Ph.D. clinical or medical geneticist should be in charge of the clinical genetics unit. Furthermore, at least half of the staff of any such unit who are directly involved in patient care, evaluation, or counseling should be board certified or board qualified.
2. Written records on all evaluations, counseling sessions, and testing done on patients/families by the clin-

ical genetics unit should be prepared. In most cases written documentation should be sent to the referring and/or private physician(s), referring agency, and/or other appropriate parties. Reports should be sent out in a timely fashion.

3. A follow-up letter reviewing the information presented and the counseling provided should be sent to any patient/family who has received genetics evaluation and counseling. The letter should be written in language that is understandable to the patient/family counseled and should be mailed out within a reasonable time period.

4. All patient information and records should be retained in a secured place at all times and should be locked up during nonbusiness hours. Computerized data should be accessible only to authorized personnel.

5. Medical information and/or records should be released only after receipt of written permission from the patient or parent(s)/guardian(s).

6. At an appropriate time following the provision of services to a patient/family, the patient/family-unit record should be reviewed. Criteria for the review of the record should be established in writing, and chart deficiencies or problems discovered during the evaluation or follow-up of the patient/family should be promptly corrected.

7. Monthly or bimonthly unit meetings should be held to discuss functional activities and to resolve issues within the unit.

8. Services by the clinical genetics unit should not be denied on the basis of age, gender, race, color, creed, or national origin.

9. Adequate and appropriate space should be available for examinations, evaluations, and counseling of patients/families.

10. A system for handling patient and staff complaints should exist. Complaints should be reviewed at the monthly or bimonthly unit meeting, and appropriate decisions should be made about the complaints.

11. A mechanism that insures adequate follow-up on the evaluation and testing that has been done on patients should exist.

The following additional guidelines are recommended for each clinical genetics unit:

1. Patient records and radiographs should be maintained indefinitely.
2. Patient/family privacy during evaluations and counseling sessions should be strictly maintained.
3. A referral system for patient follow-up evaluations, in either routine or emergency situations, should exist.

A network of local and national resources for patient/family referral for evaluation and laboratory testing should be identified by the clinical genetics unit.

4. For laboratory testing of patients and families, each clinical genetics unit should utilize certified laboratories or laboratories that participate in state (provincial) or federally recognized quality-control programs.

5. Each unit should determine the minimum components of the patient/family-unit record.

6. Periodic case conferences to discuss case histories and patient-related issues should occur.

7. Appropriate continuing education opportunities for the unit's staff should be available and utilized.

8. Written job descriptions of all positions in the unit should exist, and staff evaluations on each staff member should occur at least annually. The evaluation of each staff member should be discussed in private.

9. A mechanism should be in place for handling after-hours requests for information on evaluation and testing of patients.

10. Services by the clinical genetics unit should not be denied on the basis of the ability to pay.

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THE EVALUATION OF CLINICAL SERVICES SUBCOMMITTEE, GREAT LAKES REGIONAL GENETICS GROUP:

DAVID D. WEAVER,* DIANE BAKER,† MICHELLE THEOBALD,‡ FELISSA COHEN,§ RICHARD A. KING,|| SHIRLEY KRUEGER,# RENATA LAXOVA,** AND MARK S. LUBINSKY††

**Department of Medical and Molecular Genetics, Indiana University School of Medicine, Indianapolis;*
†*Department of Human Genetics, the University of Michigan Medical School, Ann Arbor;* ‡*Medical Genetics and Birth Defects Center, Henry Ford Hospital, Detroit;* §*Department of Medical-Surgical Nursing, College of Nursing, the University of Illinois at Chicago, Chicago;* ||*Division of Genetics and Metabolism, Departments of Medicine and Pediatrics, and the Institute of Human Genetics, University of Minnesota Medical School, Minneapolis;*
#*Rehabilitation Service of North Central Ohio, Mansfield;* ***Wisconsin Clinical Genetics Center, University of Wisconsin—Madison, Madison;* and ††*Genetics and Birth Defects Center, Children's Hospital, Milwaukee*

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Reevaluation of the Linkage of an Optic Atrophy Susceptibility Gene to X-chromosomal Markers in Finnish Families with Leber Hereditary Optic Neuroretinopathy (LHON)

To the Editor:

One of the commonest reasons for sudden-onset optic nerve degeneration in young men can be attributed to maternally inherited Leber hereditary optic neuroretinopathy (LHON) (Nikoskelainen et al. 1987). Specific point mutations at either np 11778 (Wallace et al. 1988) or np 3460 (Howell et al. 1991; Huoponen et al. 1991) in mitochondrial DNA (mtDNA) encoding for respiratory enzyme complex I subunits (i.e., ND4 or ND1) can be found in 70% of families. These mutations exist as being either homoplasmic or heteroplasmic, but the correlation between the degree of heteroplasmy and the risk of developing optic atrophy is far from clear (Holt et al. 1989; Vilkkii et al. 1990). Neither does heteroplasmy explain the strong male bias seen in LHON families, when the sex ratio of patients with visual impairment is observed.

Our earlier results indicated that susceptibility to optic atrophy in Finnish families with LHON was probably determined by an X-chromosomal gene closely linked to DXS7 with a maximum lod score of 2.48 at recombination fraction (θ) of 0 (Vilkkii et al. 1991). Exclusion of such a regulatory gene was previously accomplished in Tasmanian families, albeit with slightly different linkage analysis criteria than were used in Finnish studies (Chen et al. 1989). Likewise, the linkage of a putative optic atrophy liability locus to DXS7 and surrounding markers was later ruled out in British, Italian, and German families (Carvalho et al. 1992; Sweeney et al. 1992). However, using statistical calculations based on segregation analysis in LHON families,

Bu and Rotter (1991) ended up with a two-locus model supporting the existence of an X-chromosomal factor as an explanation for the male preponderance in patients with loss of vision.

Contradictory results prompted us to reevaluate the existence of an X-chromosomal visual loss susceptibility gene in Finnish LHON families. Pedigrees included in the previous linkage study were revised through clinical examinations and more extensive genealogical studies. As a result, phenotype status was corrected for some family members. Because we used stricter criteria in age limits, some individuals in the youngest generations were removed from linkage analysis. Furthermore, five new pedigrees (three with the 11778 mutation, one with the 3460 mutation, and one harboring neither of these) and previously untested family members were included in the study. To avoid the effect of possible genetic heterogeneity, families were separated in linkage analysis according to the mtDNA mutation. New markers acquired from the critical region included two hypervariable microsatellites from the MAO A and MAO B loci, both of which are tightly linked to DXS7. Because no recombinations between the MAO and DXS7 loci have been encountered, it is justified to replace the relatively uninformative DXS7 with the allele information gained from the MAO loci.

Ninety-four individuals from the seven families with the ND4 mutation were included in the linkage analysis (fig. 1). Families 1–4 are the same as those in our previous analysis. Linkage analysis was also performed in two families with the ND1 mutation (including family 5 from the previous study) and in two other families having neither the 11778 nor the 3460 mutation (including family 6 from the previous study). All except three individuals with no visual impairment had passed the age of 30 years. Primer sequences and PCR conditions for amplifying MAO A and MAO B microsatellite regions were taken from the original descriptions (Black et al. 1991; Konradi et al. 1992). The linkage data were analyzed with the program package LINKAGE (Lathrop et al. 1984), version 5.04. Two-point lod score (Z) values were calculated with the program MLINK. The criteria for defining phenotype status followed the guidelines used in the previous study; namely, only patients with optic atrophy were classified as affected. Gene frequency of .5 was set for the putative liability gene, but other values between .08 and .9 were also tested. The penetrance value of .01 for optic atrophy was assigned to heterozygous females to cover the assumed effect of X-chromosome inactivation. To minimize the amount of unknown arbitrary parameters in linkage analysis, penetrance values for three liability classes were derived