PERSPECTIVES IN GENETIC COUNSELING

NATIONAL SOCIETY OF GENETIC COUNSELORS, INC.

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RECENT BOOKS FOR GENETIC COUNSELORS

The Unborn Patient, by Michael R. Harrison, Mitchell S. Golbus, and Roy A. Filley, Orlando, Grune & Stratton, Inc., 1984, 455 pages, $59.50.

This resource book discusses in extensive detail virtually all aspects of dealing with a fetus in utero. It begins with an historical perspective of the fetus as a patient, tracing various views ranging from the "homunculus" of the 1600s up to the present view of the fetus visualized with sophisticated technology.

The book is divided into six major parts. The first deals with diagnosing fetal disorders and includes detailed descriptions of ultrasound and a short discussion of amniocentesis and fetoscopy. The ultrasound section is especially lengthy and details extensively both normal and abnormal fetal anatomy. The second part discusses selection for treatment, including medical, ethical, and legal considerations. I found this to be a very useful and concise discussion of many dilemmas involved in fetal treatment.

The third section discusses particular defects or diseases individually. Each treatable or potentially treatable malformation or metabolic disorder is accorded its own chapter. The authors use case histories, and there is an extremely detailed description of each procedural step, including methods, materials, and outcome. Defects discussed include metabolic disorders, nonimmune fetal hydrops, abdominal wall defects, diaphragmatic hernia, hydronephrosis, and hydrocephalus. Finally, the authors discuss development of future techniques and animal models currently in testing stages.

This book might have been titled *Everything You Wanted to Know About Fetal Surgery.* I found the book difficult to read at some points because of the volume of information and detailed discussions of each procedure, defect, anatomical structure, and statistic. I was somewhat disturbed by the fact that there is only a short discussion of genetic counseling as it relates to fetal treatment and that the rest of the book seems to leave the mother, father, and rest of the family by the wayside. However, this seems to be a textbook on how to do fetal diagnosis and treatment. It is intended more for perinatologists and ultrasonographers—those who deal with the treatment procedures—than for counselors or even medical geneticists who deal with the families.

The book is very enthusiastic about treatment, diagnosis, and intervention. Therefore, I was pleased to read in the conclusion the following statement: "Although fetal treatment offers new hope for the fetus with a correctable defect, the risks are high and there is considerable potential for doing harm.... Because a procedure *can* be done does not mean that it *should* be done."

I recommend this book as a supplementary reference for genetic counselors and medical geneticists involved in fetal treatment. However, I doubt that counselors in a general genetics unit or even prenatal diagnosis would find the book particularly helpful.

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Prenatal Diagnosis: Proceedings of the Eleventh Study Group of the Royal College of Obstetricians and Gynaecologists, edited by C.H. Rodeck and K.H. Nicolaides, London, John Wiley & Sons, 1984, 398 pages, $32.00.

This volume is a collection of review papers and commentaries on the present status and future of prenatal diagnosis, presented primarily by participants from the United Kingdom. It is a useful reference for all genetic counselors, whether or not they are currently working directly in prenatal diagnosis. The rapidly-changing technology has not only added to the list of disorders that can be diagnosed, but frequently alters the entire approach to the workup of an at-risk couple.

The volume begins with a clear overview of presently available techniques for prenatal diagnosis: amniocentesis, fetoscopy for blood and tissue sampling, and chorion biopsy. The indications for and risks of each procedure are discussed and presented in a concise table format. A detailed summary of recombinant DNA methodology and its application to prenatal diagnosis concludes this overview.

The section on chromosomal anomalies briefly summarizes the data on who is at risk and addresses the areas of difficulty in interpretation of unusual results. That is the least detailed section of the volume, and thus, probably the least useful for those working in prenatal diagnosis.

The sections on Mendelian disorders have several papers that describe in detail advances in the prenatal diagnosis of hemoglobinopathies, from fetal blood sampling to first trimester diagnosis by recombinant DNA techniques. One paper succinctly reviews the general approach to prenatal diagnosis of metabolic disorders using various types of tissue and includes a table of metabolic disorders presently diagnosable in the second trimester. Individual papers are devoted to advances in the prenatal diagnosis of severe combined immunodeficiency, skin disorders, cystic fibrosis, the hemophilias, and muscular dystrophies. Those in-depth presentations of the current status of prenatal diagnosis are not available in any other volume.

A highly informative set of papers focuses on neural tube defects (NTDs). An excellent discussion of maternal serum alphafetoprotein (AFP) screening and follow-up amniotic fluid AFP and acetylcholinesterase levels will be of particular interest

as MSAFP screening becomes more widespread in this country. The paper describing the expanding use of ultrasound in the diagnosis of NTDs in the United Kingdom is also particularly applicable to MSAFP screening programs. The final paper in that group is a detailed and balanced review of the data on dietary supplementation and the prevention of NTDs.

The papers on advances in ultrasound diagnosis of structural anomalies give a comprehensive review of the approach to congenital heart defects, skeletal dysplasias, gastrointestinal tract anomalies, and urinary tract anomalies. The paper on fetal therapy concisely describes the limited successes to date in dietary supplementation and drug therapy, and invasive procedures including shunting and catheterization. The paper on the postmortem examination of the fetus therapeutically aborted for fetal anomalies emphasizes the important area of follow-up and confirmation of results. That is often not included in discussions of prenatal diagnosis, but is clearly necessary to ensure a high degree of accuracy in a rapidly-changing field.

The volume ends with the final recommendations of the study group. It is encouraging to see that one recommendation is that "counseling services need expansion and of particular value would be more 'genetically trained associates' " (p. 397).

The volume reads well and is a useful reference. The limitations are 1) the papers were presented one and one-half years ago and recombinant DNA research has already made some information outdated, 2) there is some overlap of material in different papers and variability in the amount of detail presented, and 3) the commentary after the papers often addresses concerns particular to clinical genetics in the United Kingdom. Overall, however, this book is a much needed summary of the state of the art in prenatal diagnosis.

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Strategies in Genetic Counseling: Clinical Investigation Studies, edited by Beth A. Fine and Natalie W. Paul, Birth Defects: Original Article Series, Vol. 20, Number 6, White Plains, New York, March of Dimes Birth Defects Foundation, 1984, 212 pages.

The first publication of the proceedings of a conference of the National Society of Genetic Counselors (NSGC) offers genetic counselors and other readers an introduction to clinical research techniques and related statistical analyses. This multiauthored collection of contributed papers and workshops is a form of continuing professional education for genetic counselors.

The book is divided into three sections: (1) a general introduction to clinical research; (2) workshops on clinical research design and data collection; and (3) contributed papers relating to the application of clinical research planning, data collection, and interpretations of outcomes.

Of particular interest in Section I is Judith Hall's presentation of the clinical research on congenital contractures, which includes an enlightening classification and reference chart that describes limb contractures with involvement in other body areas. Dr. Hall distinguishes the various congenital contractures from the heterogeneous group of arthrogryposis and discusses recurrence risks, associated anomalies, and inheritance patterns. In this paper, and throughout the book, it is interesting to observe the different approaches to genetic counseling used by various authors. Several presentations emphasize the importance of a detailed family history in providing insights into basic mechanisms, which may lead to the recognition of new entities or to a more specific diagnosis.

Several workshops and papers address the psychological and psychosocial aspects of family situations. One paper presents a study design and a detailed discussion of counseling for normal siblings of an affected individual. Pauline Park shares a short, sensitive, group study on the psychodynamics of guilt and grief associated with pregnancy termination for genetic reasons. A workshop on the utilization of computer analysis and storage techniques for genetic practice and research is also summarized. Lowell E. Sever describes epidemiologic approaches for establishing screening studies and details concepts and methodologies. Anne L. Matthews discusses the new concept of "the fetus as a patient," and describes the impact on families of prenatal approaches to fetal diagnosis and care.

The clinical studies address many other issues such as stigma resulting from the knowledge gained through genetic counseling, feasibility of premarital testing, fears associated with amniocentesis, differences between informative and uninformative research, and many other equally interesting topics.

This book is not a must for everyone's bookshelf. However, it is a unique book for individuals about to embark on an investigational project and who have limited experience in clinical research. The book does not go into as much detail about specific genetic entities and situations as the reader might wish to explore, but clearly that was not the purpose of the conference. There is, however, a strong emphasis on psychological and social sequelae of genetic counseling and problems in family situations. That area is becoming an increasingly important part of genetic counseling in addition to the technical medical information provided for counselees.

In summary, this book offers various clinical investigation approaches and applications and is valuable as a reference for genetic counselors, social workers, psychologists, and other professionals.

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Practical Genetic Counselling, 2nd edition, by Peter S. Harper, Bristol, England, John Wright and Sons, Ltd., 1984, 336 pages, $24.00

Practical Genetic Counselling is a book full of useful information and insightful discussions. It is oriented towards clinical genetics, adopting an academic approach only when necessary to understand a clinical problem. Peter Harper has a gift for presenting complex situations in an understandable way for the clinician untrained in medical genetics.

Professor Harper's introduction contains some pearls of wisdom about genetic counseling. Especially useful are a table of

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background risks in the general population, an excellent discussion of estimation of risks, and some well-thought-out remarks on "directive" counseling. His warning that "it is important to recognize that not all risk estimates are of the same type" is an important statement for families and professionals.

The weak points of the introduction are mainly of omission. First, the definition of genetic counseling is limited in scope, although I have not yet found a definition that is not limited. Harper's definition intimates prevention of birth defects as a major theme, although elsewhere he defends the idea that service to an individual family generally takes precedence over "societal concerns." One would probably find disagreement on this point among many practitioners in the field. Secondly, Harper does not consider the role of the discussion of prognosis and anticipatory guidance, which is an important part of genetic counseling. Finally, he is unable, in the introduction and later in the chapter on the genetic counseling clinic, to incorporate adequately the role of the genetic associate (the master's-level genetic counselor) into the team approach. I suspect that largely because that role has been developed mainly in the U.S. and Canada. The lack of acknowledgment of that role perpetuates a lack of recognition in the general medical community of the genetics team approach (genetic counselor/genetic associate plus medical geneticist) and the growing independent responsibilities of the master's-level counselor, particularly in prenatal diagnosis. That team approach can help unite the diagnostic and counseling processes, which Harper feels are best not separated.

One of the most impressive things about the main body of the book is the wealth of tables, especially those containing data not easily available in reference form, such as risks for non-Mendelian disorders, or data not assembled with a viewpoint to providing genetic counseling. The book is organized into general chapters on overall components of genetic counseling and 15 chapters on specific organ systems detail specific disorders. The early chapters contain particularly excellent discussions of penetrance, expressivity, dominance, recessiveness, multifactorial inheritance, calculating the carrier frequency for recessive diseases, and an interesting method for calculating risks for non-Mendelian disorders when there is a lack of empirical data.

In the chapter on chromosome abnormalities, the discussion of risks when there is a previously affected child is not a comprehensive look at the data, but presents certain sets of data. As Hook (1) points out, the figures for the recurrence of Down syndrome provided in major clinical genetics texts are remarkably inconsistent and based on insufficient data. There is a useful table on the approximate likelihood that a Down syndrome patient whose chromosomes have not been studied represents an inherited translocation.

In the chapter on prenatal diagnosis, Harper states that prenatal diagnosis is acceptable only if pregnancy termination is being considered. He has some definite ideas about the magnitude of risk and how it should affect the patient's choices. His risks for complications of an amniocentesis are somewhat higher than many centers in the U.S. would quote. There is an excellent table on risk in twins.

Harper's discussion of consanguinity in the chapter on special problems in genetic counseling is excellent, but newer data do not support the stated lack of increased risk in consanguinous unions in populations with a long tradition of consanguinous marriage (2, 3). His warning comments on dysmorphology are extremely well-taken.

In general, the chapters on specific diseases, organized by organ system, are good; they are full of useful risk figures, although some of them could be better referenced. One potential pitfall is that this information changes rapidly. Counseling for rare conditions, medical management for common conditions, and prenatal diagnosis change so rapidly, in fact, that reliance on any single information source will quickly result in the use of outdated information and failure to maintain the best standard of care.

The book is extremely useful as a reference in conjunction with updated reviews of the current literature. *Practical Genetic Counselling* is also helpful for professionals in practice who are trying to interpret information relayed by specialists in medical genetics and trying to prepare their patients for what to expect when more specialized counseling of diagnosis is needed. The book ably fulfills the author's stated purpose: "making an appreciable number of clinicians aware of the importance, scope, limitations and pitfalls of genetic counseling in their own work."

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CORRESPONDENCE

To the Editor:

When patients come for counseling due to exposure to low doses of x-rays or nonteratogenic drugs, simple "good news" counseling can take on a confusing twist. I have personally experienced a transition in style and then returned to the original style when doing such counseling. I wish to share the failure of my short-lived style.

Originally and currently, the sequence for a counseling session for nonteratogenic environmental exposure would be as follows:

1. introduce myself
2. ask why the patient is there
3. indicate that I will speak to the patient's concerns, but first need to ask questions
4. take the pedigree
5. get details of environmental exposure
6. explain concepts of teratogenesis: timing, quantity, and teratogenicity of particular substance
7. state that the substance in question did not likely increase risks

I had been using this method frequently, when I thought it might be better to relieve the patient's anxiety right at the beginning of the counseling session. Why not, for example, state #7 right after #1 and #2: "Mr. and Mrs. Jones, I will get into it in more detail later, but let me start by saying that as far as we can tell, the substances you were exposed to will not increase your risk." That is, rather than have the patient worry throughout the session until I say all is okay, reveal the good news at the beginning.

The problem is, that while this approach sounds reasonable, it

does not work. Each time I have tried this method of presenting the good news up front, in the patient's eye, my credibility seemingly went out the window. Apparently, such a simple answer is not expected at that point. I hypothesize that there is a need for a gradual development in presenting that type of good news. A simple, up-front statement of "no increased risks" seems to catch the patients off guard, causing disbelief and skepticism.

I have gone back to my original method of gradually unfolding the "story," and ending with "... so, therefore, we do not believe that this substance will increase the risks." Patients once again seem satisfied, content, and in full belief of the conclusion having seen its development.

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CASE REPORTS IN GENETIC COUNSELING

Response to Case #4 (Vol. 7, No. 1, March 1985)

I read with great interest the case report describing two situations in which couples presented for chorionic villus sampling with seemingly high genetic risks, although no medical records were available to substantiate their histories. In both cases, upon discussing the test results and providing subsequent follow-up with those couples, the counselor became suspicious about the couples' true motivation for requesting parental diagnosis. Both of the couples appeared to demonstrate inappropriate concern about the sex of their infants.

In her report, Ms. Baum presents an excellent analysis of the ethical issues and emotional considerations that health-care providers must confront when serving families who choose to use, or sometimes misuse, accessible methods of prenatal diagnosis for sex selection. However, Ms. Baum did not address the possibility that cultural factors may have influenced the behavior of those couples. In both cases the couples were from cultures different from that of the counselor—one being Indian and the other Pakistani. It is human nature for us unintentionally to project our value system to others such that we assume our customs and beliefs to be universal. This phenomenon often leads to barriers in genetic counseling as cultural relativity begins to influence our perceptions of how patients ought to behave. For example, when dealing with a family from another culture in the context of perinatal genetics, it is important to be knowledgeable about their specific beliefs and customs concerning gender, birth order, congenital malformations, and neonatal death.

To help genetic counselors become acquainted with cultural behaviors during the childbearing cycle and explore strategies for dealing with those issues with families, an additional workshop has been scheduled for the upcoming NSGC National Education Conference in Salt Lake City, Utah. (This workshop did not appear in the list of concurrent workshops outlined in the meeting brochure.) Together with Vicki Colburn, a clinical educator in obstretrical nursing, I am preparing a workshop titled "Behavior During the Childbearing Cycle: A Cultural Potpourri." One of our primary goals is to engender a better understanding of cultural issues in order to enhance the effectiveness of health care we can provide to families from other cultures. We are especially excited about the opportunity to address genetic counseling issues relevant to perinatal care and look forward to learning from you, our colleagues.

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POSITIONS AVAILABLE

Genetic Counselor: The Genetics & IVF Institute of Fairfax, Virginia, is seeking to employ a second full-time genetic counselor. The Genetics & IVF Institute has an established prenatal genetics program in both amniocentesis and chorionic villus sampling, with an increased volume of pediatric-adult genetics referrals. The institute staff is representative of the following specialty areas: medical genetics, biochemical genetics, cytogenetics, genetic counseling, obstetrics, pediatrics, nursing, *in vitro* fertilization, and reproductive endocrinology. Members of the staff are encouraged to seek academic appointments or research affiliations with one of the major institutions in the area. The National Institutes of Health, Medical College of Virginia, and Georgetown University are represented among current staff appointments. Responsibilities will include genetic counseling for prenatal and pediatric patients, implementation and provision of genetic services in the public health setting, teaching in professional and community education programs, and assisting with clinical procedures. The position requires a master's degree in genetic counseling or related field and board certification/eligibility by the American Board of Medical Genetics. Nursing experience is desirable, but not essential. Interested candidates should send a curriculum vitae and names of two references to: Shirley L. Jones, RN, MS, Genetics & IVF Institute, 3020 Javier Road, Fairfax, VA 22031, phone: (703) 698-4355.

Genetic Associate: A position is available 1 December 1985 at the clinical faculty (assistant in pediatrics) level within the Division of Genetics, Department of Pediatrics at the University of Florida. A master's degree in genetic counseling and certification by the American Board of Medical Genetics or eligibility are required. This is a diversified position with involvement in genetic counseling for prenatal diagnosis and a wide range of disorders in a pediatric population, education of health professionals, and participation in satellite and specialty clinics and clinical research. Interested applicants should submit a curriculum vitae and three letters of recommendation by 1 November 1985 to: J.L. Frias, MD, Professor and Chief, Division of Genetics, Box J-296, JHMHC, University of Florida, Gainesville, FL 32610. For more information about the position, please call Shearon Roberts or Sonja Rasmussen at (904) 392-4104. The University of Florida is an equal opportunity/affirmative action employer.

Regional Coordinator – MSAFP Program: A position is available immediately for a master's-level RN or genetic counselor to serve as regional coordinator for Northern California Kaiser-Permanente maternal serum alphafetoprotein screening program, Oakland. The coordinator will supervise the screening of 25,000 pregnant women per year for fetal neural tube defects and chromosomal anomalies. Responsibilities will include monitoring flow of patients and laboratory results; supervising patient education; maintenance of liaison with laboratory, physicians, genetics units,

and the State of California Department of Health; coordinating in-service education of health-care providers; compilation of program statistics; and counseling individual patients. Experience in administration and patient education or counseling is preferred. The position provides full-time clerical support. Salary negotiable based on experience. Contact: Bruce Blumberg, MD, 2200 O'Farrell Street, San Francisco, CA 94115, phone: (415) 929-5059; or Ronald Bachman, MD, 280 West MacArthur Blvd., Oakland, CA 94611, phone: (415) 428-5964.

Genetics Counselor: The Prenatal Diagnosis Center of Harbor-UCLA Medical Center has a full-time, permanent position available as of 1 January 1986 (possibly sooner). Primary responsibilities involve pre-amniocentesis and CVS counseling, as well as other issues in prenatal diagnosis and reproduction. The Prenatal Diagnosis Center is part of a large, comprehensive genetics division at Harbor-UCLA, with many educational opportunities. Salary commensurate with experience. Please send curriculum vitae with references to: Nicki Chun, MS, or M.M. Kaback, MD, Division of Medical Genetics, Building E4, Harbor-UCLA Medical Center, Torrance, CA 90509, phone: (213) 533-3759.

Genetics Counselor: The Department of Human Genetics at Yale University School of Medicine is seeking a genetic counselor to function in the daily operation of the Prenatal Diagnostic Unit. Responsibilities will include performing counseling and obtaining medical histories of patients undergoing amniocentesis, chorionic villus sampling, and fetoscopy. Duties will also include the reporting and interpretation of abnormal maternal serum alpha-fetoprotein (AFP) levels to obstetricians and arranging for follow-up care when indicated. Participation in the development of clinical findings and research efforts of the Prenatal Diagnostic Unit is also required. Contact: Miriam Schoenfeld, MSW, Supervising Genetic Counselor, Prenatal Diagnostic Unit, Department of Human Genetics, Yale University School of Medicine, I-130 SHM, P.O. Box 3333, New Haven, CT 06510-8005, phone: (203) 785-2667.

Genetic Associate: A new genetic associate position is available in Medical Genetics, University of British Columbia, to commence immediately. Formal genetic associate training preferred (MS degree), practical experience desired. Send curriculum vitae and names of three references to: J.G. Hall, MD, Medical Genetics, Grace Hospital, 4490 Oak Street, Vancouver, B.C., V6H 3V5. U.B.C. is an equal opportunity employer. Preference will be given to Canadian citizens or landed immigrants.

Genetics Counselor: A position is available for a master's-level genetic counselor who is board eligible or certified. The job involves primarily prenatal counseling for maternal age. We have the only level-three ultrasound unit in a large metropolitan area, and therefore see quite a few major congenital anomalies prior to birth. We will expect our counselor to be involved in postnatal parental counseling involving neonatal deaths and still-births, especially where a congenital anomaly is involved. We are currently doing about 250 amniocenteses per year. Starting salary will range between $20,000 and $25,000, depending on qualifications and experience. Contact: Donald L. Levy, MD, Director, Division of Maternal-Fetal Medicine, Childrens Hospital of the Kings Daughters, 800 West Olney Road, Norfolk, VA 23507, phone: (804) 628-3976.

JOBS HOT-LINE
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Genetic Counselor: The University of Utah Medical Center has an immediate opening for a full-time genetic counselor. Responsibilities include direct services in the areas of general genetics, prenatal diagnosis, specialty clinics, and outreach clinics. Involvement in community education and clinical research is encouraged. The successful applicant will work with three other genetic counselors with the support of three medical geneticists. Master's degree and board certification or eligibility required. Salary commensurate with experience. The Rocky Mountain region offers many outdoor amenities including skiing, camping, and hiking. Send curriculum vitae to Janice C. Palumbos, MS, Genetic Counselor, Department of Pediatrics, CRW 413, University of Utah Medical Center, Salt Lake City, UT 84132, phone: (801) 581-8943. An equal opportunity employer.

Genetic Associate: Genetic Associates of North Carolina, Inc. has an opening for a full-time genetic associate. Applicants should be board certified/eligible as a genetic counselor with the American Board of Medical Genetics. Preference will be given to applicants who have at least two years of work experience as a genetic associate. Responsibilities will include general and prenatal genetics clinic services. Salary commensurate with background and experience. Please send curriculum vitae and references to: Philip D. Buchanan, PhD, Genetic Associates of North Carolina, Inc., Suite 107, 104 South Estes Drive, Chapel Hill, NC 27514, phone: (919) 942-0021. Genetic Associates of North Carolina, Inc. is an equal opportunity employer.

ANNOUNCEMENTS

Region VI Conference

The next NSGC Region VI Conference will be held at Asilomar, California, 19-22 March 1986. For further information, please contact Beth Crawford, Genetic Counseling Option, Bldg. T-7, University of California, Berkeley, CA 94720.

Longitudinal Study of Infants Exposed to Accutane

Maternal use of isotretinoin (Accutane) during the first trimester has been associated with a high risk for major malformations and probably an increased risk for miscarriage. The Massachusetts General Hospital is conducting a study to assess the risks for other adverse outcomes among infants exposed to Accutane during pregnancy, including minor malformation, hearing and visual deficits, and immune deficiency. Participating families would not incur expenses from the study and would not be asked to travel; all evaluations would be performed locally. We are also interested in referrals of Accutane-exposed fetuses. It would be optimal if exposed fetuses were reported as early in gestation as possible (i.e., before any diagnostic ultrasound procedures). In addition, we are interested in receiving pathologic results, or arranging for pathologic evaluations, of electively or spontaneously aborted Accutane-exposed fetuses. Anyone with information concerning Accutane-exposed fetuses or infants is requested to contact: Edward Lammer, MD, Embryology-Teratology Unit, Massachusetts General Hospital, Boston, MA 02114, phone: (617) 726-1742.