PERSPECTIVES IN GENETIC COUNSELING

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PATERNITY TESTING: AN OVERVIEW

Edward M. Kloza, Glenn Palomaki, Richard Mahoney, and Thomas G. Brewster.

The biblical account of King Solomon settling a dispute concerning parentage by threatening to divide the child in two is one of the earliest recorded attempts at using a medical-legal solution to such a problem (1).

Fortunately, the application of the Mendelian laws of inheritance to the ABO blood group system provided the first scientific basis for the resolution of questions about paternity. The characterization of other red blood cell antigens and, most recently, the characterization of the human leukocyte antigen (HLA) system, have given the determination of paternity a scientific basis. However, the interpretation of paternity test results often utilizes a Bayesian approach by integrating objective test results with assumptions about prior probability of paternity.

Background

A paternity test may be initiated by an individual request or by a court order. Several situations (2) may prompt the need for this type of information: inheritance claims, child custody disputes, amendment of birth records, welfare compensation, naturalization claims, and child support. Child support is currently one of the most common reasons for requesting paternity testing. Public Law 93-647, passed by Congress in 1975, requires each state to develop an appropriate plan for the ascertainment of paternity and for enforcement of child support.

The selection of a gene system for use in paternity testing is dependent on the following characteristics (2):

1. There must be a single and unequivocal pattern of inheritance.
2. Phenotypes must be accurately classified by reliable techniques.
3. The system should be unaffected by environmental factors.
4. The common alleles should have relatively high frequencies.

In any case involving disputed paternity, one first attempts to demonstrate that the tested male is *not* the father. Laboratories that rely soley on testing red blood cell antigens are able to exclude up to 70 percent of falsely accused males. The HLA system, because it is highly polymorphic, is the single most powerful commonly used system in paternity determination; its use can exclude 90 percent of falsely accused males. Testing for both red cell antigens and HLA types can effectively exclude more than 96 percent of falsely accused males. The addition of red cell or serum enzyme testing (2) or chromosome heteromorphism analysis (3) would increase the probability of exclusion to well above 99 percent. When the alleged father is not excluded, this testing is able to provide information about the likelihood or plausibility of paternity.

Interpreting Paternity Test Results

Complete interpretation of paternity test results requires consideration of four factors:

1. the alleged father's prior probability of fathering the child,
2. the laboratory's average cumulative probability of exclusion (CPE), based upon the number and type of genetic markers tested,
3. the paternity index (PI), and
4. the individualized CPE (ICPE) of the mother and child.

1. Prior Probability

The prior probability of fathering the child is simply the likelihood that the alleged father is the biological father. This probability is based on information and evidence other than test results derived from the examination of genetic markers. For example, the courts will often consider the question of access, that is, the likelihood that the alleged father had sexual relations with the mother at a time consistent with the conception of the child. If the alleged father can prove that he was out of state or otherwise inaccessible during that critical period, his prior probability would be very low. Conversely, if it were shown that he and the child's mother were living together during that time, the prior probability may be considerably higher. In either case, the laboratory is never in a position to determine the prior probability of paternity and should neither solicit such information nor consider it when reporting test results.

2. Average Cumulative Probability of Exclusion (CPE)

When a decision is made to have a paternity test done, one chould choose a laboratory with a high average CPE. Because the average CPE represents the percentage of falsely accused males excluded by testing, such a choice will maximize both the probability of correctly classifying a falsely accused male and the ability to estimate the plausibility of paternity if no exclusion is found.

Some testing systems have a higher power of discrimination than others. Therefore, CPEs will vary among laboratories, depending on the systems used. Laboraties testing the same genetic markers should have the same CPE. To determine the CPE for a given laboratory, the probability of exclusion (PE) for each individual gene system tested must first be determined. This depends on the number of alleles in the system, the gene frequencies of the alleles, and the number of alleles tested for by the laboratory.

Consider the Kell and Kidd systems, both of which incorporate two alleles (2). The frequencies of the two alleles of the Kell (k and K) and Kidd (Jk^a and Jk^b) systems in the Caucasian population are p(k) = .964, q(K) = .036, and p (JK^a) = .510, q(JK^b) = .490. The probability of exclusion for a two allele system in which two alleles are detected is calculated by using the formula pq(1-pq). Therefore, the PE is .033 for the Kell system and .187 for the Kidd system. Even though they are both two-allele systems, testing for both Kidd alleles will exclude 18.7 percent of falsely accused random males, while the Kell system excludes only 3.3 percent because the frequencies of the two Kidd alleles are more evenly distributed. However, because anti-JK^b reagents are scarce, a laboratory may test only for the JK^a antigen, in which case the appropriate formula for calculating the PE is pq^4, resulting in a PE of .029. A PE of .029 indicates that 2.9 percent of random males would be excluded as the biological father based on testing of the Kidd JK^a allele only, a six-fold reduction compared to the results obtained if both Kidd alleles were tested.

To combine the PE from several tests one must first calculate the probability of inclusion, that is, the probability that the alleged father will be included by *all* of these tests. Then the cumulative probability of exclusion (CPE) is equal to one minus the cumulative probability of inclusion.

The table below lists the tests typically performed by paternity testing laboratories and demonstrates how the addition of test systems to the protocol increases the likelihood of excluding a falsely accused male.

Test	Markers Tested	PE	CPE
ABO	A, B	.145	.145
Kell	K, k	.033	.173
Duffy	Fya, Fyb	.181	.323
P	P1	.027	.341
MNS	MS, Ms, NS, Ns	.316	.549
Rh	C, c, D, c, E, e, C^w, D^u	.320	.694
HLA	*	.891	.967

* At least 20 A alleles and 40 B alleles are tested.

3. Paternity Index (PI)

When no exclusion is found, one can determine a paternity index (PI). The PI describes how many times more likely it is the alleged father produces sperm that carry the genes consistent with fathering the child than does a random male from a similar population. This, of course, assumes that the mother is the biological mother.

The example below uses the ABO system to determine the PI. Standard blood typing techniques can identify four phenotypes: A, B, AB, and O. Consider the following situation:

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Individual	Phenotype	Genotype
Child	A	AA or *AO*
Mother	B	BB or *BO*
Alleged father	AB	AB*
Biological father	?	AA, AB, or AO

*The genotype of the alleged father cannot, of course, b determined with certainty for phenotypes other than AB or O.

The obligatory gene, that is, the gene that the biological father is obliged to contribute, can be identified. If the mother is the child's biological mother, her genotype must be BO and the child's AO. Therefore, the obligatory gene is gene A. Because the alleged father is type AB, he is not excluded; furthermore, 50 percent of his children will inherit gene A.

The frequency of gene A in the Caucasian population (.2738) is used to calculate the PI:

$$PI = \frac{\text{Probability of alleged father contributing obligatory gene}}{\text{Probability of random male giving obligatory gene}}$$

$$= \frac{.5000}{.2738} = 1.826$$

Based solely on the results of ABO typing, the alleged father is 1.826 times more likely than a random male to be genetically capable of fathering the child.

4. Individualized Cumulative Probability of Exclusion (ICPE)

When the testing is completed and the results analyzed, the laboratory may report, in addition to the PI, the individualized CPE, which describes how many males in the appropriate population would be excluded as the biological father based solely on the phenotypes of the mother and child. Thus, if the mother and child have common phenotypes the ICPE may b low. On the other hand, if the child has a rare phenotype, th ICPE would be much higher. To calculate the ICPE for a mother and child one must first determine individual probabilities of exclusion (IPE) for each genetic system tested and then combine those probabilities. The IPE of the ABO system for the mother and child in the example used earlier is the probability of being a phenotype that does not include the obligatory allele A, that is, phenotypes B or O.

ICPE = Probability of being phenotype B + probability of being phenotype 0 = .0912 + .4361 = .5273

The IPE figures are derived from the following phenotypic frequencies for the Caucasian population:

A = .4366
B = .0912
O = .4361
AB = .0361
1.0000

Because the ICPE is .5273, 53 percent of the population of Caucasian males would be excluded as the biological father of this child by this mother. The probability could also be expressed as the number of random males who would need to be tested to find one who could produce sperm capable of fathering the child in question. In this example, 2.1 men would need to be tested in order to find one with the obligatory gene.

Determination of Plausibility of Paternity

Note that the laboratory's contribution involves the determination of factors 2, 3, and 4. The establishment of the prior probability and how it is to be integrated with the paternity

index is outside the realm of the laboratory. However, it is instructive to see how it may be used by the courts.

For purposes of interpreting test results useful in a court of law, a prior probability of .5 is assumed. That is, the laboratory assumes that in the absence of test data, the alleged father is just as likely as not to be the biological father. If a different prior probability is considered, its modification by the test results using Bayes Theorem will redefine the plausibility of paternity. Only by using the prior probability of paternity and the PI can one give an estimate of the odds of paternity. For example, with a PI of 19, depending on the prior probability, the odds of paternity can vary from 2:1 to more than 170:1, given prior probabilities of .1 and .9, respectively. In this example, a PI of 19 and a prior probability of .5 would translate into a plausability of paternity of 19:1, or 95 percent (19/20).

Communication of Test Results

When the alleged father is excluded by paternity testing, results are unequivocal and should be clearly and concisely stated as such. When no exclusion is found, all parties to the action must understand that such a finding never carries with it any certainty that the alleged father is the biological father. However, it is reasonable, in the presentation of results, to place the probability of paternity in its proper perspective.

Just as a couple at risk for having a child with a recessive condition may perceive a one in four risk as being vastly different from a 25 percent risk, a jury may perceive a 95 percent probability of paternity as being different from a likelihood of paternity of 19:1. Furthermore, although a jury may consider a 99.4 percent probability of paternity to be only slightly stronger than a 95 percent probability, there is almost a 13-fold difference, because the respective likelihoods are 241:1 versus 19:1.

Summary

Maintenance of services for paternity testing involves many elements associated with traditional genetic counseling activities such as understanding modes of expression of a variety of multi-allelic systems, statistical analysis of test results using Bayesian probability, and the effective communication of likelihood estimates in a manner that is most useful to the consumer.

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THE GENETIC COUNSELOR AND CHORIONIC VILLI SAMPLING

Dorothy M. Halperin

The recent development of chorionic villi sampling (CVS), a first trimester method of prenatal diagnosis, provides a new option for many couples at risk for chromosomal or biochemical disorders. It also provides an exciting opportunity for the genetic counselor to become an integral member of the CVS team. At Michael Reese Hospital and Medical Center, the first U.S. medical center to do CVS on continuing pregnancies, we have been actively involved in providing genetic counseling to candidates for this procedure and in coordinating our CVS program.

CVS is a prenatal diagnostic procedure that is done at 8 to 10 weeks after the last menstrual period. It involves the passage of a plastic catheter with a metal obturator through the vagina and cervix and into the uterus. Under ultrasound guidance, the catheter is positioned in the chorion frondosum, where, utilizing negative pressure, a small sample of this rapidly dividing tissue is aspirated. The chorionic villi can be used for chromosomal or biochemical analyses; due to the nature of the villi, results of these studies are available in 1 to 10 days.

Genetic Counseling

Genetic counseling is an absolute prerequisite to chorionic villi sampling. The purpose of the counseling is to inform patients thoroughly about the procedure, so that they will have correct and complete information on which to base a decision about using CVS for their pregnancies. Genetic counseling is done at least one day prior to CVS, although we prefer to counsel patients earlier. We provide basic information about CVS and answer patients' questions by telephone prior to a formal genetic counseling session. Some acceptable CVS candidates decide against the procedure on the basis of the telephone discussions.

We counsel the CVS patients in a small group setting and try to limit these groups to four patients or couples. Following the group meeting, which takes about one hour, we see each patient individually to discuss her particular indication for CVS and her specific genetic risks, to obtain a pedigree or family history, and to answer any questions she may wish to ask privately.

During the group discussion, we compare CVS to amniocentesis and emphasize that the latter is the low-risk standard of care. Both procedures are defined and described, and the benefits and disadvantages of each are highlighted. Disadvantages of amniocentesis include the fact that is is done at 15 to 16 weeks and requires a 3- to 4-week wait for results. Many patients perceive the needle as a negative feature. In addition, many are concerned that abnormal results require a second trimester termination of pregnancy as the only way to avoid the birth of an affected child. The benefits of amniocentesis include the low procedural risk and the fact that there is an attendant, routine assay for alphafetoprotein (AFP) to screen for open neural tube defects.

Attractive features of CVS are that it is done at 8 to 10 weeks, that no needle is involved, that results are available in one day with a direct preparation and are confirmed in 7 to 10 days with short-term tissue culture, and that the patient can elect a first trimester pregnancy termination if an abnormality is diagnosed. The drawbacks include the fact that the risks of CVS cannot be quantified at present—although we do know that the types of complications are similar to those of amniocentesis, for example, spotting, infection, leakage, and spontaneous abortion. In addition, the overall accuracy of the CVS analysis is unknown.

During the group session we tell the patients exactly what to expect before, during, and after CVS. We stress that this is a team effort and describe the role of each team member. We inform the patients that they will hear the team interaction during the procedure, including some potentially upsetting comments such as "stop" or "I can't see the catheter;" we explain the meaning of these comments.

We describe the procedure as uncomfortable, but not painful. The discomfort is due to the full bladder and the use of a tenaculum, or tissue clamp, which is placed on the cervix. We show the patients a sample catheter and share with them diagrams of the catheter passage and photographs of 8- to 10-week-old fetuses, depicting the appearance and location of the chorionic villi. Chorionic villi are defined as immature or pre-placental tissue. We mentioned that only a very small portion of this tissue is necessary for genetic analysis and explain that the geneticist will examine the tissue under a microscope to ensure that a sufficient quantity of villi has been obtained. We inform the patient that a second pass of the catheter will be attempted if there is insufficient tissue. We explain that in some cases samples cannot be obtained due to the length of the catheter (at 21 cm it may be too short) or a physical/anatomical blockage. If sampling is unsuccessful, the patient is offered an amniocentesis at 15 to 16 weeks.

We define chromosomes for the patients and explain our methods for direct and short-term tissue culture. We inform the patients that they will be notified by telephone of the results of the direct preparation for chromosomes (primarily chromosome number), and that these results are considered preliminary until confirmed by short-term tissue culture (chromosome number and structure). We emphasize that this testing is not 100 percent accurate and discuss the problems posed by laboratory error, mosaicism, and maternal cell contamination, which also occur with amniocentesis. Results of biochemical tests are communicated to the patient as soon as they become available. The time for completion of those analyses varies with the specific assay used; for example, results for Tay-Sachs disease (TSD) are usually available within hours, whereas DNA restriction analysis can take up to two weeks. We stress that neither CVS nor amniocentesis can rule out every type of genetic anomaly, birth defect, or mental retardation.

During the group genetic counseling session we discuss thoroughly our experience to date and provide information about the number of patients sampled, loss rate following the procedure, instances of infection, number of babies born, number of abnormal results, and male:female ratio. We do not quantify the risk of the procedure because our sample is still small, the number of successfully completed pregnancies is even smaller, and our series is uncontrolled. A percentage based on our data may, therefore, be misleading at this early stage.

We present CVS from the perspective of risk vs. benefit, where an unknown procedural risk is weighed against one's numerical risk for and burden of the genetic problem in question, and aginst the benefits of the procedure. We do not try to coerce patients into accepting or declining CVS. There is, of course, no right or wrong decision about CVS; the patients are encouraged to do what feels right and comfortable for them. All CVS candidates are told that some form of prenatal diagnosis, either amniocentesis or CVS, is medically indicated for their pregnancies.

On leaving the group meeting the patients are given a packet of materials that includes a written description of CVS, the three-page consent form and information about our follow-up protocol. The protocol involves ultrasound evaluations at 12 and 16 weeks; screening of maternal serum AFP at 16 weeks (to compensate for the fact that CVS does not routinely screen for open neural tube defects); and provision of information about the remainder of the pregnancy, labor, and delivery, and examination of the newborn.

We do not routinely recommend amniocentesis for CVS patients. Certain patients at risk for biochemical disorders, however, have had confirming amniocenteses, particularly when they were the first to have CVS for the disorder in question. Once a diagnosis of abnormality has been made on chorionic villi, confirming amniocentesis is no longer necessary. Our patients at risk for fragile X syndrome and Fanconi anemia have had amniocenteses that confirmed a diagnosis of the absence of the disorders based on CVS. One could also recommend amniocentesis in cases of mosaicism found in chorionic villi samples. If the pregnancy is electively terminated due to the diagnosis of a genetic abnormality, confirmation of the diagnosis and a pathologists's examination of the fetus and the placenta are requested. If a pregnancy is lost spontaneously following CVS, we request the date and circumstances and, if possible, have a pathologist examine the fetus and placenta.

Patient Attitudes

To date, most of our CVS patients have been self-referred; many are more familiar with this procedure than are their obstetricians. The majority of patients who actually come for genetic counseling are highly motivated and are, for the most part, already committed to having CVS. As a group, they seem to be less concerned with the risk of the procedure and appear less anxious than amniocentesis patients. We feel that this may be due to the fact that CVS does not involve a needle, and that many patients therefore erroneously assume that CVS carries no risk at all, or less risk than amniocentesis. Some patients, however, opt against CVS following genetic counseling primarily due to the fact that its risk are unknown.

We note with interest that many women find second trimester amniocentesis unacceptable since the advent of CVS. Now that there is an alternative, many women are unwilling to wait until 15 to 16 weeks for the test, and another 3 to 4 weeks for results. They are also unwilling to undergo a second trimester pregnancy termination, a procedure that is more difficult medically and psychologically than a first trimester termination. Patients also feel that first trimester CVS protects the privacy of the pregnancy until the results of the genetic studies are completed. With amniocentesis, on the other hand, the pregnancy is often very obvious prior to obtaining results.

Organizational Issues

The use of CVS poses some practical genetic counseling and screening dilemmas. For example, we routinely recommend carrier testing for TSD, sickle cell anemia, and thalassemia for Jewish, black, and Mediterranean couples respectively, prior to amniocentesis. It may be very difficult to obtain these carrier tests prior to CVS, because time is often a limiting factor. This is a particular concern for TSD, where a pregnant patient or couple may require a white-cell analysis. When a couple is identified as carriers of thalassemia, it often takes several weeks to complete the DNA restriction analyses that will determine whether prenatal diagnosis is possible. CVS also allows less time to follow up on any pedigree information that may indicate that a couple is at additional reproductive risk.

Since CVS involves a team—an obstetrician, ultrasonographer, clinical geneticist, cytogeneticist, and a genetic counselor—it is extremely important that the team work well together and be available on a regular basis for CVS. Our team is available to do the procedure three days per week, and genetic counseling can be done as needed. It is a good idea to have more than one person who can function in each role. Because this is an early procedure, it often must be scheduled quickly. This has not been a major problem for us, because most patients comply with obtaining the necessary information (ultrasound, GC culture, blood type) and come to us at the appropriate time.

Conclusion

Our group genetic counseling for CVS, coupled with short individual meetings, appears to be working well. We inform patients of this arrangement prior to their arriving for genetic counseling; none has voiced objection to this format. Many have said that the group experience was especially helpful, because others asked questions that they had not thought of. Positive comments from the patients confirm our conviction that genetic counseling is an extremely important component of chorionic villi sampling.

Medical centers throughout the country are preparing or have begun to offer CVS, and there is an increased number of patient requests for the procedure. The genetic counselor will play a vital role in the design and implementation of comprehensive, high quality programs for chorionic villi sampling, and in the education of health-care professionals and the public about this new technology.

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CASE REPORTS IN GENETIC COUNSELING

Note: This is the first in a continuing series of case reports in genetic counseling. We encourage all professionals involved in providing genetic services to submit cases of interest and to comment on the cases published here. Please see Perspectives *Vol. 5, No. 4, December 1983 for specific instructions on preparing case reports.*

The consultand was a 27 year old, G4,P3,Ab0 female seen for amniocentesis counseling because of the prior delivery of an infant with Down syndrome (47, XY +21). She decided to have the amniocentesis, and results were 46 XX, normal female. After the patient was informed of these results, she repeatedly called back to inquire about the likelihood of error regarding the sex of the fetus, to make sure her husband was informed by us of the fetus' sex, and, finally, to obtain her ultrasound results so she could have a termination performed.

We referred her to the radiology department because we were unsure of the policy on releasing ultrasound information to the patient. (She said she was no longer seeing the physician who had referred her to us.) The ultrasonographer who was present during the aminiocentesis later informed us that what appeared to be uppermost in the patient's mind during the tap was the patient's strong desire to have a boy. In addition to the boy with Down syndrome, the patient had two normal girls, and she stated repeatedly that if this fetus were female, the couple would keep trying until they "got a boy."

Because the patient had no phone, we were unable to call her and ask her why she was terminating this pregnancy. Had we been able to reach her, we are not sure how we could have avoided sounding as if we were prying.

The current concern of our staff is this: Because the patient has stated her strong desire to have a boy, we are reasonably certain that we will see her again should another pregnancy occur. Because of the previous delivery of the child with Down syndrome, she is justified in seeking an amniocentesis. However, we are uncomfortable in dealing with her due to our strong suspicions that this pregnancy was terminated because of sex.

Our options include:

1. referral to another center,
2. seeing her again, but discussing our suspicions and concerns over her "misuse of resources,"
3. seeing her and treating her as any other patient (that is, ignore the sex issue), and
4. seeing her, but refusing to inform her of the fetus' sex.

Total objectivity would require our acknowledging that what this woman does with her own body and her fetus is her concern alone. However, it is often difficult to maintain such an objective approach; our humanity and previous experiences interfere. For example, during the week that this situation occurred, we also saw a woman whose third pregnancy was diagnosed as Down syndrome (her first pregnancy was also diagnosed as Down syndrome, the second ended in miscarriage) and a woman whose fourth pregnancy was diagnosed as anencephalic (the first ended in stillbirth, the second, in a first trimester pregnancy loss, and the third, in an anenephalic fetus). The juxtaposition of these cases was ironic and seemed to emphasize the unfairness of the situation. In addition, our feminist tendencies made us resent the implication that a girl was less desirable than a boy, although the reverse situation would be no more palatable to us.

We would like to resolve our somewhat hostile feelings about this patient so that we are able to deal with her fairly should she return to our clinic. We are interested in hearing about similar situations and methods of dealing with them.

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CORRESPONDENCE

To the Editor:

I am just starting a private practice in genetic counseling and would like to pass on to the NSGC the information I have gathered with respect to professional liability insurance.

Goode & Webster has been offering a policy for some time. The cost is $750 for $1 million coverage (Goode & Webster, The Market Place, Manlius, NY 13104).

St. Paul Insurance Company has indicated a cost of $413 per year for $1 million coverage. There are St. Paul agents in most large cities, or one can contact Peter S. Postma at J. Braniff & Co., One Greenway Plaza East, Suite 700, Houston, TX 77046, phone: (713) 623-2330.

A group policy that seems to be very good is available through the Fred S. James & Co. of Texas, Inc. (350 Glenborough #100, Houston, TX 77067) to those who join one of the divisions of the American Association for Counseling and Development (AACD). I joined the American Mental Health Counselors Association (AMHCA) division. The annual premium is $37.50 for $1 million coverage. In addition, the policy provides an attorney and pays legal fees in the event of litigation.

The AACD has more than 40,000 members. AMHCA has 7,000-8,000 members in a variety of counseling disciplines. I talked with the president of AMHCA, and he enthusiastically welcomes all interested genetic counselors into the association. For additional information about AMHCA call: 1-800-354-2008. For membership information write: American Association for Counseling and Development, 5999 Stevenson Avenue, Alexandria, VA 22304, phone: (703) 823-9800.

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Neural Tube Defects; Amniocentesis for Prenatal Diagnosis of Genetic Disorders; and *Genetic Disorders.* American College of Obstetricians and Gynecologists, 1983.

The American College of Obstetricians and Gynecologists (ACOG) publishes a flyer listing its patient education pamphlets. The three titles under review deal directly with genetic counseling issues.

Neural Tube Defects summarizes the availability of prenatal testing for spina bifida, provides definitions of spina bifida — the "open spine" form and the "closed spine" form, and describes anencephaly. Tests for maternal serum and amniotic fluid alphafetoprotein (AFP) are described in detail, and the pamphlet states clearly that the tests are not foolproof. Although 50 out of 1000 women have elevated maternal serum APP, only one to two carries a fetus with a neural tube defect. The pamphlet urges individuals to discuss this testing with their physicians, spouses, and counselors. The authors note several times that although there is an increased risk for those who have a family member with a neural tube defect, 95 percent of couples who have a child with spina bifida have no personal or family history of the disorder. One flaw in this publication is that it does not state who is a candidate for AFP screening. The authors note, of course, that anyone with a family history should have this test, but they do not recommend that other groups be tested, despite the prior assertion that most children born with these defects are born to families without a positive family history. The pamphlet is concise and does an excellent job of explaining neural tube defects, as well as the testing that is available. The authors recommend that ultrasound and amniocentesis follow a determination of elevated serum AFP. This sequence may not be standard procedure in all centers. In general, however, this pamphlet is very useful for those wishing to maximize the chances of a healthy pregnancy outcome.

The pamphlet *Amniocentesis* is quite complete. There is a discussion of birth defects in general, a brief discussion of chromosomes, and a discussion of the types of abnormalities that can be detected by amniocentesis. The procedure is described in detail, and the pamphlet states that the results of amniocentesis are available in three to four weeks. Chromosome analysis and the various tests for AFP are described. The authors mention that fetal sex determination is especially helpful in "detecting sex-linked disorders such as muscular dystrophy." This statement can be misleading to those who are not aware that there is no prenatal test for Duchenne muscular dystrophy.

The risks of amniocentesis are reviewed in detail. A paragraph at the end of the pamphlet emphasizes the need to discuss the facts and the couple's feelings when abnormal results are found. This pamphlet is also concise and well-developed. For a short flyer, it is very useful, although it lacks the graphic pictures of chromosomes and other aspects of prenatal diagnosis that are seen in other, longer publications.

Genetic Disorders is a short pamphlet that briefly reviews basic cell biology, chromosomal abnormalities, Mendelian inheritance, and multifactorial inheritance. There is a nicely written section on genetic counseling and how it can benefit couples. The authors also discuss prenatal testing and newborn screening. The pamphlet is generally reassuring; the author states, for example, that "some 93 percent of all children are born healthy, with no significant birth defects." The author urges couples to be well informed about their risks and to discuss their plans with their physician so that the proper medical decisions can be made.

These three pamphlets are quite useful because they are short, to the point, and contain quite a bit of information. They may be useful in the genetics clinic, but would also be beneficial at local health fairs or in the waiting rooms of private physicians so that people can be made aware of the availability of genetic services. These pamphlets are available from the ACOG Distribution Center, 600 Maryland Avenue, S.W., Suite 300 East, Washington, DC 20024-2588. The minimum order is 100 copies. The cost is $20 for 100 pamphlets, $75 for 500 pamphlets (which can be made up of a mixture of a number of different pamphlets), and $150 for 1000 pamphlets. The price is higher for shipment to Canada, Mexico, and other countries. One may also order the complete directory *Patient Education Pamphlets.*

The Needs of Children with Spina Bifida: A Comprehensive View. Mark Wolraich, 1983.

This 35-page booklet can be extremely useful in the spina bifida clinical setting. The purpose of the book is to emphasize the broad-based and multidisciplinary needs of children with spina bifida. The author advocates a multidisciplinary clinic that deals with the "whole child." The booklet emphasizes "normalization" of the disabled child and the importance of a positive experience with professionals in helping new parents of a child with spina bifida cope with their child's condition.

The booklet opens with a case report of a baby born with spina bifida. There is a detailed description of the etiology and incidence of spina bifida, as well as the terminology used in neural tube defects: meningocele, myelomeningocele, spina bifida cystica, spina bifida occulta, and myelodysplasia. The booklet reviews in depth the effects of spina bifida on the various body systems and describes how these medical problems can be managed. Again, a case history is used to illustrate the effectiveness of working within a spina bifida clinic. The service and counseling needs of the family are discussed in detail. The section titled "Adjustment Tasks Facing Families" is useful as a refresher for most genetic counselors. There is some discussion of education programs for children with these disabilities. Genetics is discussed very briefly, but, unfortunately, the list of service providers for the generic spina bifida team does not include a geneticist or genetic counselor.

The third section of the booklet discusses the use of primary and tertiary care for these families, as well as sources of funding that vary from state to state. The book is useful for parents who are involved in such a system because it can allow them to keep track of the professionals they should be seeing and will alert them to at least some of the services available to them. If parents do not live near a multidisciplinary, university-based clinic, this information can help ensure they set up appointments with appropriate private-practice physicians and other professionals.

From a clinical standpoint, the booklet is useful in that it emphasizes the psychosocial aspects that surround the patient and family when a birth defect occurs. Much of the policy information could be left out of a family book, but it is informative to medical students and other professionals who may be new to this type of system. The booklet is available by writing to: Division of Developmental Disabilities, Department of Pediatrics, University Hospital School, The University of Iowa Hospitals and Clinics, The University of Iowa, Iowa City, IA 52242.

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MEETINGS AND ANNOUNCEMENTS

Annual March of Dimes Symposium

The Seventh Annual New York March of Dimes Symposium on Genetics for the Practicing Physician will be held Sunday, 23 September 1984 at the Grand Hyatt - New York. The title of this year's symposium is "Clinical Application of the New Genetics." The symposium is presented by the Greater New York March of Dimes and the Genetics Task Force of New York and is open to all interested health professionals. Approval is pending for six credit hours, Category I, CME. For additional information, contact: Leona J. Schumer, March of Dimes, 622 Third Avenue, New York, NY 10017, phone: (212) 922-1460.

APHA Genetics Committee Established

In response to the increasing number of maternal and child health issues related to clinical genetics and genetic counseling, the American Public Health Association (APHA), at its 1983 annual meeting, initiated the establishment of a genetics committee. The committee is chaired by Raymond Kessel, coordinator of Wisconsin Statewide Genetics Services. Among the initial objectives are recruitment of genetics professionals to the APHA; planning of genetics sessions in APHA annual meetings; and provision of a forum for leadership in determining the role of genetics, research priorities in genetics, and allocation of resources in order to make "genetic services available to all members of society in a public health model."

The committee sponsored its first event on 16 November 1983 at the APHA annual meeting in Dallas, when it held a joint session with the Public Health Nursing section; the session was titled "Issues in Genetic Counseling and Screening." The committee met later that day to hear a report on the federal genetic diseases program. The report was presented by Allan S. Noonan, chief of the Genetic Disease Services Branch. Following the report, the committee discussed additional plans and objectives. For additional information, contact: Raymond Kessel, Coordinator, Statewide Genetics Services Network, 104 Genetics Building, 445 Henry Mall, Madison, WI 53706, phone: (608) 263-6355.

Maine Enacts Genetics Legislation

On 11 May 1984, the state of Maine passed L.D. 2099, "An Act to Amend the Statutes Relating to Handicapping Conditions Under the Human Services Law." This bill, which becomes law on 25 July, establishes and funds a voluntary statewide genetics program under the auspices of the Department of Human Services. The program offers testing, counseling, and education to parents and prospective parents. In the past, the provision of clinical genetic services to residents of Maine has been contingent upon the awarding of federal grant monies to the Department of Human Services for various public and private agencies.

Correction

The March 1984 issue of *Perspectives* listed an incorrect phone number for information about the 1985 NSGC education conference to be held 7, 8 October in Salt Lake City, Utah. Information can be obtained from Barbara Biesecker, conference chairperson, at: (608) 263-1991 or 262-1006.

POSITIONS AVAILABLE

Genetic Associates: Two new positions are available for genetic associates in medical genetics, University of British Columbia, to commence immediately. The positions involve collection of precounseling data, counseling of prenatal and specialty clinic cases, the screening of referrals, and follow-up of families. Public education involvement and participation in research projects required. Formal training as a genetic associate preferred (i.e., MS degree), practical experience desired. Send C.V. and three references to: Dr. J. Hall, Medical Genetics, Grace Hospital, 4490 Oak St., Vancouver, B.C., V6H 3V5. The University of British Columbia offers equal opportunity for employment to qualified male and female candidates. Preference will be given to Canadian citizens or Landed Immigrants.

JOBS HOT-LINE NUMBER
Linda Nicholson: (302) 651-4234

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