

The DNA Files
Prenatal Testing: Do You Really Want To Know Your Baby's Future?
Transcript

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Prenatal Testing: Do You Really Want To Know Your Baby's Future? Transcript

JOHN HOCKENBERRY: This is the DNA Files, I'm John Hockenberry. [*baby cry*]
Every parent wants a healthy baby.

JUDY: I was about 17 weeks pregnant and knew that I was supposed to get an amnio.
And then I thought, you know, no one in my family ever had a baby with a genetic
problem.

CLAIRE SHERMAN: I was extremely nervous about the test causing miscarriage, but I
was even more nervous about the possibility of Down's Syndrome. I had the feeling that
that was the one thing I really couldn't deal with.

JOHN HOCKENBERRY: Prenatal tests allow us to know more about the health of our
unborn babies than ever before, but do we know enough to make life or death decisions?
Should individuals or society decide what we mean by "healthy baby"? For the next hour
the DNA Files will explore Prenatal Genetic Testing: Do You Really Want To Know Your
Baby's Future?

But first:

JOHN HOCKENBERRY: Well before a child is born doctors can peer into its genetic
inheritance. Prenatal testing can now identify over three hundred genetic traits, some of
them crippling disorders. But when the new tests go wrong, doctors and laboratories face
a new kind of lawsuit, one that argues that the child should never have been born. John
Rieger has this report.

JOHN RIEGER: She's a little girl named Savannah, and by all accounts she's a sweet,
loving child. But in 1997 her mother had to tell a Texas court that she should never have
been born. Savannah was born with spina bifida, a disabling deformation of the spinal
cord, and her parents sued their doctor, not because he caused the disability but because he
didn't detect it in time for an abortion. It's called "wrongful birth".

MARSHA SAXTON: The implications of the wrongful birth suit are that Savannah's
birth was a mistake, and that the quality of her life would be so terrible that the only
compensation would be millions of dollars for her parents.

JOHN RIEGER: Marsha Saxton also has spina bifida. She's married, has a Ph.D, and
holds positions at the UC Berkeley School of Public Health and the World Institute on
Disabilities. Though disabled activists are often at odds with the medical establishment,
Saxton calls wrongful birth suits against doctors and hospitals cruel and misguided.

MARSHA SAXTON: The legal system forces people in lawsuits to present their case in
the strongest terms. Parents who've had a chance to bond with a child, to get to know that
child a bit, will almost inevitably love that child. And their lawyers have to coach them to
present their lives as burdened by the child. That in and of itself profoundly distorts the
parent-child relationship with those families.

JOHN RIEGER: Birth defects have always been one of the risks of pregnancy. So, how did they become a legal cause of action? Prenatal tests can now discover all kinds of disorders in the unborn fetus. Genetic tests for sickle cell, thalassemia, Down's Syndrome and Tay Sachs Disease are routine procedures for some populations. These disorders can bring real suffering to those affected and their families. And a majority of pregnant women, when informed their fetus has a serious incurable genetic disease, choose to have an abortion. But everything depends on the tests. If the hospital makes a mistake, the consequences to the family can be severe.

DR. BERTRAM LUBIN: The family was told that the results were normal, but that was a mistake. They sent it for the wrong test.

JOHN RIEGER: Dr. Bertram Lubin is director of medical research at the Children's Hospital in Oakland. Dr. Lubin recently testified for the plaintiff in a wrongful birth suit involving a disabling genetic blood disease.

DR. BERTRAM LUBIN: This is a family who had already had a child with thalassemia trait, and their next child had thalassemia disease. Um...it's a tragic mistake for the child who's going to now have to be transfused for life, and chelated later on. So this is bad, in my opinion, and I do think somebody is responsible.

JOHN RIEGER: In cases like this parents have successfully sued doctors and hospitals for the extra cost of raising their disabled child, which may amount to millions of dollars by the time of adulthood. But some plaintiffs have gone further, arguing that it's the child who deserves to be compensated for expenses that will last a lifetime. Geneticist and attorney Pilar Ossorio of the Institute for Ethics at the American Medical Association says these so-called wrongful life suits have foundered in the courts.

PILAR OSSORIO: That child is basically claiming that but for the negligence of this doctor of this lab, I would not have been born. So, the harm that the child is claiming is their very existence. And that's something that the court has found incredibly difficult to deal with.

MARSHA SAXTON: Children are born wanting to be alive.

JOHN RIEGER: Disabilities activist Marsha Saxton.

MARSHA SAXTON: It's impossible to determine the burden to one's self of one's own life, and to then try to compensate for that so-called loss by virtue of one's existence. You know the obvious remedy would be to kill that person if they are such a burden to themselves.

JOHN RIEGER: While twenty-two states allow wrongful birth suits, only three – California, Washington State and New Jersey – allow wrongful life suits. Instead of untangling the philosophical questions, says Pilar Ossorio, these states simply ask, Who should pay?

PILAR OSSORIO: What the court says is – they say, well, we think it's most

appropriately put on the doctor or the doctor's insurance, and so that's basically what they're doing. They're making an economic decision.

JOHN RIEGER: Wrongful birth and wrongful life lawsuits are a disturbing byproduct of prenatal testing and a healthcare system that often relies on the courts to apportion the costs of long-term care.

PILAR OSSORIO: The whole thing is, if nothing else, incredibly distasteful. You know, what we need is a healthcare system that takes care of people with disabilities.

JOHN RIEGER: For the DNA Files, I'm John Rieger.

JOHN HOCKENBERRY: This is the DNA Files. I'm John Hockenberry.

[*Newborn shrieks*] Isn't he beautiful!

JOHN HOCKENBERRY: A new baby...for every parent a window on a lifetime of unknowns. What will this baby become? Parents have asked that question since the beginning of time. But today they can ask some new questions. Besides simply counting on fingers and toes, many modern pregnancies are guided by an often complex, high tech process that can screen for hundreds, and some day perhaps thousands of attributes.

GENETIC COUNSELOR: This is saying we're going to draw the maternal serum alpha-fetoprotein test, and it's a routine screening, because you don't have a history of a previous neural tube defect and your age isn't very old, your gestational age today is 16 weeks and 2 days.

JOHN HOCKENBERRY: This simple blood test is one of a series of prenatal screening tools that doctors use to try to look into your baby's future.

RUTH SCHWARTZ COWAN: People have been interested in some kind of prenatal diagnosis for all time.

JOHN HOCKENBERRY: Professor Ruth Schwartz Cowan

RUTH SCHWARTZ COWAN: You probably know the various folk tales and folklore – if you're carrying high it's a boy, if you're carrying low it's a girl, and if you're carrying to one side it's going to have brown hair, if you're carrying to the other side it's gonna have black hair. Those are all forms of prenatal diagnosis, but what we're talking about basically has its start in the post World War II era.

JOHN HOCKENBERRY: Ruth Schwartz Cowan is an historian of science and technology at the State University of New York at Stonybrook.

RUTH SCHWARTZ COWAN: As hard as it is for us to believe this today, back in the 30s and 40s it was impossible to see human chromosomes under the microscope clearly.

JOHN HOCKENBERRY: But once they could, another discovery soon followed. A French physician who worked with mentally retarded children discovered that most of his patients had a chromosomal abnormality in common. Again, Dr. Schwartz Cowan.

RUTH SCHWARTZ COWAN: It is the most common form of Down's Syndrome. Now, it occurred to medical geneticists that if they could prenatally diagnose Down's Syndrome, and they knew what the chromosomes looked like, the question was, could you get fetal cells so you could look at them and determine whether or not a fetus had Down's Syndrome?

JOHN HOCKENBERRY: While geneticists were working in the late 50s and early 60s to find a way to grow living cultures from the fetal cells they could extract from a pregnant woman's womb, outside of these laboratories several events occurred which brought the taboo topic of abortion into the public arena.

CYNTHIA GORNEY: The first was a very famous incident that took place in 1962.

JOHN HOCKENBERRY: Cynthia Gorney's book "Articles Of Faith, A Frontline History Of The Abortion Wars", includes a profile of a young Arizona woman named Sherry Finkbein. Pregnant with her fifth child, and suffering mightily from nausea, Finkbein swallowed a supposedly mild tranquilizer her husband had brought back from Europe. Soon after, Sherry Finkbein saw news reports about a drug called thalidomide, and took the pills to her doctor.

CYNTHIA GORNEY: He took a look at what it was, went and looked it up in his manual – and said, I have very bad news for you, this contains thalidomide, there's a very good chance that the baby you are carrying is severely deformed. If you were my wife, I would urge you to end this and start over again.

JOHN HOCKENBERRY: But in 1962, abortion was illegal, except in cases in which the mother's life would be endangered by having a baby. Still, says Gorney, there was an unspoken system back then. Her doctor could arrange for the procedure discreetly, for some vague health reason, without ever using the A word. Such was the plan for Sherry Finkbein, until a local newspaper ran a story about the plight of an unnamed local woman who'd taken a baby-deforming drug. Panicked hospital officials cancelled Finkbein's procedure.

CYNTHIA GORNEY: And within about two weeks this was a national story trumpeted all over the United States – it was something everybody was talking about, and a tremendous debate began, really for the first time in American social history, for the previous forty years – should this woman be permitted to have this abortion or not?

JOHN HOCKENBERRY: Sherry Finkbein traveled to Sweden to have the abortion. Then, in the mid-60s, a rubella epidemic swept through the United States. Pregnant women exposed to rubella faced a 50% chance of bearing a child with severe deformities. Once again, some physicians arranged for their patients to have abortions.

[Pro-choice chant in background]

JOHN HOCKENBERRY: While the cultural war over legalizing abortion raged, scientists were quietly engaged in clinical trials of a prenatal genetic test called amniocentesis. They'd figured out how to grow little cell cultures and then check to see if the chromosomal mutation that indicated Down's Syndrome, called trisomy 21, was present. In 1973, the United States Supreme Court announced its landmark decision in Roe v. Wade, which legalized abortion in the first trimester of pregnancy.

[Pledge of Allegiance by anti-abortion demonstrators]

JOHN HOCKENBERRY: By 1975, researchers in Canada, Great Britain and the United States had concluded their studies of the impact of amniocentesis on pregnant women and their fetuses. Professor Ruth Schwartz Cowan.

RUTH SCHWARTZ COWAN: It was determined that the procedure was relatively safe. The miscarriage rate was acceptably small, and that's when amnio started to become standard practice, at least in the developed countries of the west.

JOHN HOCKENBERRY: Fear of wrongful birth lawsuits, filed by parents who had not been offered testing and then gave birth to a baby with a disability, convinced doctors that offering prenatal genetic testing was a form of professional insurance. Lori B. Andrews is a professor at Chicago Kent College of Law.

LORI B. ANDREWS: I think that the biggest thing that lead to the dispersion of prenatal genetic testing technology were lawsuits by patients who hadn't been offered screening. For example, women over age 35 who hadn't been offered the test for Down's Syndrome. So that meant that genetic testing moved very rapidly into the obstetrical community.

JOHN HOCKENBERRY: Today, it is standard operating procedure for doctors to offer prenatal genetic testing to any pregnant woman who is 35 years of age or older, or to women of any age who have a known family history of inherited conditions. Producer Kathy McAnally visited several women who had to ask themselves if they wanted to be tested to know something about their baby's future, even if that knowledge forced them to choose between having an abortion or bearing a child that might have physical or mental limitations.

KATHY McANALLY: Judy and Clair are friends. Both live in Berkeley, California, both became pregnant when they were past age thirty-five. Clair is married, and had been trying hard for a successful pregnancy for some time. The thought of prenatal genetic testing terrified her. Judy got pregnant kind of by accident. But Judy and her boyfriend were happy about the baby. Judy didn't really give the idea of having an amniocentesis much serious thought.

JUDY: I was 17 weeks pregnant and knew that I was supposed to get an amnio. And then I thought, no one in my family had ever had, you know, a baby with a genetic problem. And I thought, maybe I won't do this.

KATHY McANALLY: But, said her boyfriend, probably you should.

JUDY: And I went in with him and they showed us a film which I don't think I paid much attention to, talking about how people decide what to do with the information they get.

KATHY McANALLY: Clair Sherman was very serious and worried about her amnio. The couple had already suffered through three miscarriages, and knew that the more invasive kinds of prenatal tests could increase her chance of losing this baby.

CLAIRE SHERMAN: I was extremely nervous about the test causing miscarriage, but I was even more nervous about the possibility of Down's Syndrome. I had the feeling that that was the one thing I really couldn't deal with.

[Tour of testing lab]

KATHY McANALLY: There are several different kinds of prenatal tests. The most common is ultrasonography, which shows a rather fuzzy, indistinct image of the fetus.

LEE FALLON: You're still limited by the fact that you're not looking directly at a fetus, you're getting an indirect image.

KATHY McANALLY: Lee Fallon is a genetic counselor at the Genetics and IVF Institute in Fairfax, Virginia. Ultrasound isn't an invasive test, the image of the fetus is picked up on an external monitoring device. Another simple test is what's called the alpha-fetoprotein test. In A.F.P. a blood test measures the levels of several substances that are made by the developing fetus. Those measurements can tell doctors if the fetus shows an increased risk for having either a neural tube defect, or Down's Syndrome. It's a screening test, not a diagnostic one, says Lee Fallon.

LEE FALLON: About 5% of all women who are pregnant would screen positive. However of those 5%, about 97% of the pregnancies are likely going to be normal.

KATHY McANALLY: Remember, Fallon uses the word "likely", since none of these tests are 100% accurate. But if a woman does screen positive, or if she's 35 or older, most doctors will recommend additional testing. And these tests are more risky, and could involve putting the developing fetus at a slightly higher risk of miscarriage.

LEE FALLON: The two most common diagnostic tests that are used in the United States are amniocentesis and chorionic villus sampling. Amniocentesis has been around for about 28 years and involves taking a relatively small amount, about a tablespoon, of the amniotic fluid out, using a very thin needle that, with the ultrasound on, is guided into the sac where the fetus is.

KATHY McANALLY: Amniocentesis is a diagnostic test, because it allows a look at the fetal chromosomes. You can actually see whether or not they are normal. With CVS, a small amount of the tissue that forms the placenta is removed, again by inserting a needle through a pregnant women's abdomen to remove the cells.

LEE FALLON: And again those cells are fetal cells, and again would give you a source to the chromosomes and the DNA and can be used again for Down's Syndrome or for other DNA-type testing.

KATHY McANALLY: Claire remembers worrying over the various testing options with a genetic counselor.

CLAIRE SHERMAN: We discussed the pros and cons of early amnio or CVS, both of which I had already ruled out because they have a higher risk of miscarriage, and with this pregnancy I had a lot of bleeding in the first trimester, every time I bled they called it a threatened miscarriage. But then I had to get through the amnio.

[Claire reading to child]

KATHY McANALLY: Claire's story had a happy ending: she's now the mother of a four-year-old daughter and is expecting her second child. In fact, on the day I visited, Claire had received an early morning call from her genetic counselor.

CLAIRE SHERMAN: I had just gotten up, and she said she was sorry if she had woken me up but she figured for good news that would be OK, and I said, absolutely. And she told me everything was fine.

KATHY McANALLY: Judy also remembers an early morning phone call. She remembers the counselor telling her...

JUDY: I have some information to give you about your amnio. And at that point I knew something was wrong. And I said, well, what are you talking about? And she said well...and she proceeded to speak and I didn't hear a word she said.

KATHY McANALLY: The amniocentesis showed that Judy's fetus had a chromosomal abnormality.

DR. DAVID WITT: Well, when we're talking about chromosomal problems, you have to realize that chromosomes are relatively large pieces of genetic information.

KATHY McANALLY: Dr. David Witt directs a medical genetics program for the Kaiser Permanente Health Plan.

DR. DAVID WITT: And any one chromosome contains thousands of genes. And when you realize that a single gene can cause a severe disease, if you consider that with a chromosome problem you're talking about literally hundreds of genes being abnormal, you can imagine the outcome can be quite serious.

KATHY McANALLY: Judy and her boyfriend were tested to see if the chromosomal abnormality could be linked to either of them. Both tested negative. And the abnormality that showed up in the cells of the fetus didn't absolutely mean that the baby would suffer from mental retardation, or other physical problems. Judy agonized over what she should do.

JUDY: I felt almost poisoned inside – that I had this information and if I gave birth to a child that was severely disabled, having had this prior information, I don't know how I would have dealt with it.

JOHN HOCKENBERRY: A dilemma living inside her. Later in this program, Judy talks about her decision.

[Musical break]

JOHN HOCKENBERRY: Do you really want to know your baby's future? Is it even possible? Lori B. Andrews, a law professor who specializes in the legal and ethical questions raised by genetic testing, says no, you can't.

LORI B. ANDREWS: There are over 4,000 genetic disorders. Even if you did every prenatal test available, that would be about 350 disorders, and it would be prohibitively expensive, so generally obstetricians only test for a subset of disorders they think the woman might be at risk for.

JOHN HOCKENBERRY: Things such as maternal age, or a family history of a certain disease. But, remember, Judy didn't have a family history of genetically linked disease, but still the tests turned up a problem. Dr. Michael Kaback is a Professor of Pediatrics and Reproductive Medicine at the University of California in San Diego.

DR. MICHAEL KABACK: It's very important for everyone to appreciate that no human being is genetically perfect. We are all carriers of multiple genetic alterations, mutations, if you want to call them. Some of those mutations may be of no consequence, some may be of substantial consequence later in our lives, some may be of substantial consequence early in our lives.

DR. DAVID WITT: Well, every living thing has a genetic blueprint.

JOHN HOCKENBERRY: Dr. David Witt.

DR. DAVID WITT: All the genes compiled together are like a set of instructions that determine how the human body grows and develops. It determines simple things like eye color, but also every other component of the body and how it works. That genetic blueprint is packaged in every cell in the body, and the blueprint is identical from one cell to the next. And it's packaged in things we call chromosomes, which are big enough to see under the microscope when you prepare them in a certain way. And then smaller units within the chromosomes called genes.

JOHN HOCKENBERRY: We all have 23 pairs of chromosomes in each cell, 22 numbered ones, with the 23rd being the pair that determines our sex. Each chromosome is formed from a long ladder of DNA. Again, Dr. David Witt.

DR. DAVID WITT: You inherit one half of the pair from your mother and one half of the pair from your father.

JOHN HOCKENBERRY: There are mathematical ratios that govern dominant and recessive inheritance. In real life, real families, these tidy mathematical formulas can translate into heartbreak.

[*Music*]

Tay-Sachs disease is a fatal neurological disorder caused by the lack of an enzyme called hexosaminidase A. In order for a child to be born with the disease, both parents must be carriers of the Tay-Sachs gene. In that case there is a one-in-four chance of affliction. In the orthodox Jewish community, which has one of the highest birthrates in America, prevention efforts have focused exclusively on pre-marital testing, as Jon Kalish reports.

JON KALISH: As any parent of a Tay-Sachs child will tell you, the onset of the disease is heart-wrenching. There are signs of delayed development at around six months of age, accompanied by the loss of muscle tone and failing vision. At 12 to 18 months the baby will have difficulty swallowing and suffer from convulsions and seizures. As pediatrician Michael Kaback puts it, these children are out of contact with their environment.

DR. MICHAEL KABACK: Ultimately, by two or two and a half years of age they get into a totally disabled state, and depending on how well they're cared for or whether or not some intervening events occur like infection or what have you, they will generally live in this sort of a chronic vegetative state, as it's called, for three or four years before they die. And they all do die.

JON KALISH: In 1971 a test on amniotic fluid became available. Prenatal testing for Tay-Sachs began in the Washington-Baltimore area and quickly spread throughout North America, Israel, South Africa and Australia. A blood test to detect carriers soon followed, and by 1985 DNA testing was added to the screening program. One in every 300 people carries the gene for the disease, with higher than average incidence amongst French-Canadians. The highest carrier rate is among Ashkenazy Jews of East European descent, where it is estimated to be as high as one in 25. The rate among the Orthodox Ashkenazy is believed to be a bit higher. Prenatal testing initially targeted married couples, who were advised to have an abortion and to practice birth control if positive. Both are anathema to the orthodox, who have a booming birthrate and a history of in-breeding. Outside of Israel, the largest concentration of Orthodox Jews is in Brooklyn, where a Tay-Sachs program provides subsidized screening.

[*doorbell chimes, patient buzzed in, door shuts*]

CLERK: Good afternoon, can I help you?

YITZY: Yes, I'm here for Tay-Sachs testing, please.

CLERK: You mean the Dor Yeshorim test?

YITZY: Um...yeah.

CLERK: OK. [*over p.a. system*] Guys, I need a technician for Dor Yeshorim. I need a technician upstairs for Dor Yeshorim. Thank you.

JON KALISH: At the modern diagnostics laboratory in Brooklyn, 26-year-old Yitzzy arrives on a sweltering weekday afternoon to have blood drawn.

TECHNICIAN: Okay, just keep your arms still for one minute. [*grunt*] There we go. Okay, bend your arm up. I'll put a band-aid on in one minute.

JON KALISH: For orthodox Ashkenazi Jews like Yitzzy, the Tay-Sachs blood test has become a rite of passage into young adulthood.

YITZY: Basically, just to make my mother-in-law happy. I mean, I'm engaged already, and I guess it's a good thing to do also, to be on the safe side in terms of diseases or what have you, that could be lurking somewhere. You never know.

JON KALISH: Pre-marital screening in the orthodox community is a unique approach to coping with Tay-Sachs, and one that is consistent with traditional Jewish law. Orthodox Jews are diligent about fulfilling the Old Testament dictate to be fruitful and multiply. And so to avoid unions which are unable to start a family, the screening program encourages young people to get tested before a relationship gets serious. The pre-marital approach is far more desirable than prenatal because abortion is largely viewed as a violation of the commandment Thou shall not kill. The procedure is, in rare circumstances, permitted for the mother's physical or emotional well-being. It is said that after the development of the prenatal test, no non-orthodox Jewish couple had a second Tay-Sachs baby. But for many orthodox families there were multiple births and then deaths of Tay-Sachs babies.

RABBI JOSEPH EKSTEIN: After I lost my fourth child to Tay-Sachs, and went through together with my wife all that suffering, I decided that something has to be done to stop this happening again in our community.

JON KALISH: And so in 1983 Brooklyn, Rabbi Joseph Ekstein founded the Association of an Upright Generation, or Chevra Dor Yeshorim as it's known in Hebrew. At his request, leading orthodox rabbis in New York issued a public call in 1986 for couples to be tested for Tay-Sachs before they marry. Rabbi Ekstein recalls there was widespread concern at the time that carriers might be stigmatized. The relatives of carriers were the most agitated about screening.

RABBI JOSEPH EKSTEIN: Those families that had the problems with Tay-Sachs, they were in tremendous fear that I am taking out the problem to the streets. They were very, very nervous about it that this was going to damage them. Even my own wife was totally against the entire idea.

JON KALISH: Orthodox rabbinical leaders directed Ekstein to develop a screening modality that would maximize anonymity and be totally voluntary. Since its inception, Dor Yeshorim has used grants and contributions to subsidize its screening program. Of the \$300 lab charge, people who are being tested pay a little over \$100. To date 90,000 have been tested and 200 at-risk couples have been identified. A date of birth, telephone number and identification number are used for records, but no names are taken. When a couple calls in to find out if a match is genetically compatible, Dor Yeshorim will only release results if both people are carriers. Rabbi Ekstein says that a tiny percentage of the

at-risk couples continue their relationships, knowing full well of the risk. Over the years several couples registered with Dor Yeshorim had Tay-Sachs children. Rabbi Joseph Ekstein says his is an odd vocation: putting an end to romances in order to prevent enormous suffering.

RABBI JOSEPH EKSTEIN: Well, let's break the relationship. It's not easy, we know. But on the other hand, how can you compare the pain of seeing a child passing away in your own hands? How can you compare one with the other?

JON KALISH: Dor Yeshorim says the number of babies born with Tay-Sachs disease in the orthodox community here went from an average of 4 or 5 a year in the 1980s to between zero and two a year today. One hospital in Brooklyn with a 16-bed Tay-Sachs ward had a waiting list, but the ward was completely closed several years ago. The California Tay-Sachs disease prevention program reports that for the larger North American Ashkenazy community the number of Tay-Sachs babies born annually has plummeted from 50 or 60 a year to between four and six. Though Dor Yeshorim started out testing just for Tay-Sachs, the organization has expanded its successful screening program to include canavan disease, fanconia anemia type C, and cystic fibrosis.

JOHN HOCKENBERRY: One of the issues Jon Kalish raised in his story is the expansion in the kinds of tests offered by Dor Yeshorim. But not all genetically linked diseases are equal in their severity. Some, like Tay-Sachs, are always fatal. Other diseases and conditions have more varied outcomes. Dr. Michael Kaback.

DR. MICHAEL KABACK: In jest, I've said to Rabbi Ekstein, if he waits long enough, he will be able to prevent every marriage in that community, since everyone is at risk for something in their offspring.

JOHN HOCKENBERRY: Later in this program, we'll visit a family living with another genetically linked disease – cystic fibrosis. This is the DNA Files.

[*Music*]

JOHN HOCKENBERRY: This is the DNA Files. I'm John Hockenberry. Like Tay-Sachs disease, cystic fibrosis is recessive, so it takes two copies of the CF mutation, one inherited from each parent, for the disease to occur. Very often, there is no family history of the disease. Rather, it can show up when one perfectly healthy carrier has children with another person with the mutation. Here again is producer Kathy McAnally.

KATHY McANALLY: He doesn't look at all sick. And while he's a bit small, Brendan charges through life armed with the same enthusiasms as other boys his age.

BRENDAN HARRIGAN: My name is Brendan Harrigan. I'm eleven years old. And I like baseball, football and basketball.

KATHY McANALLY: What do you know about cystic fibrosis?

BRENDAN HARRIGAN: Well, it makes you have a lot of mucus in your lungs and you have to take a lot of pills to digest your food.

KATHY McANALLY: Brendan was the Harrigan's third child. When he was about 8 months old, his parents noticed that he was wheezing a lot. At first he was treated for asthma. His mother, Susie, recalls that her baby kept getting sicker, and finally ended up in the hospital, inside an oxygen tent.

SUSIE HARRIGAN: And the doctor did a few tests for other problems he thought it could be, and said there was a final test he wanted to do, that he said hopefully it wouldn't be the case. And he mentioned that it was cystic fibrosis. I had never heard of CF before and had no idea what it even meant.

KATHY McANALLY: With cystic fibrosis, a protein is produced that leads to a dangerous increase in the level of salt and potassium in the body. Mucus builds up in the lungs, and digestive passageways can become obstructed. Back when Brendan was diagnosed, doctors told the Harrigans that he'd be lucky to see his 20th birthday.

SUSIE HARRIGAN: It was a real scary time for us, and we've had some scary times since then. I think we've been fortunate that when I've gotten scared or frustrated or angry, my husband's strong. And when he's gotten frightened or upset, then I've been at a good place, and fortunately too, ever since Brendan was diagnosed, and was put on medication and treated for the CF, he's never been healthier.

[Blow, Brendan, blow, blow, blow... good job!]

KATHY McANALLY: Every three months, Brendan Harrigan is evaluated by Dr. Nancy Lewis, a specialist at Children's Hospital in Oakland, California. On this day, Dr. Lewis is a little bit concerned, Brendan hasn't gained much weight since his last check up.

[Lewis and Brendan talk back and forth in examining room; she tells him he has to eat more rich food]

SUSIE HARRIGAN: I go to the store and I look at the labels. Everyone else is looking for non-fat and low fat, and I'm like, no, not enough fat, and put it back. Don't they have anything fattening anymore? Just give me some fat...

KATHY McANALLY: Brendan also uses a device daily to help clear the mucus from his lungs.

BRENDAN HARRIGAN: A flutter is this little tube with a marble in it and it shakes my chest, so it makes the mucus loose inside so I can cough that up.

KATHY McANALLY: How often do you have to do that?

BRENDAN HARRIGAN: Twice a day.

KATHY McANALLY: Do you ever get frustrated or mad that you have to go through this all the time?

BRENDAN HARRIGAN: Yup.

KATHY McANALLY: What do you say about it?

BRENDAN HARRIGAN: It stinks

KATHY McANALLY: Brendan and his family have watched the life expectancy for people with cystic fibrosis edge up, from about 20 to age 31 over the past 11 years. When Susie was pregnant with Brendan, there was no prenatal genetic test for CF. After much debate in the medical genetics community, CF carrier screening is about to be offered to all women of childbearing age, so long as appropriate counseling services are also available to those tested. Susie Harrigan.

SUSIE HARRIGAN: You know, I've sort of thought about that and I vacillate in my feelings. I used to think what you don't know can't hurt you. We had no idea. I had a great pregnancy, he was born. He looked healthy, he looked great. Nothing was wrong and I didn't have to worry. Now as I see all the things that they can do, I do think, if I got pregnant again, I'd be afraid, but I'd want to know.

[*Music*]

JOHN HOCKENBERRY: Once again it comes down to the question: Do you really want to know your baby's future? And what would you do with that information if a prenatal genetic test indicated a problem? Let's look for a moment at screening programs for cystic fibrosis.

DR. DAVID WITT: In 1989, when the gene was discovered, the whole question of population screening was raised, simply because it's a very common gene to be carried, particularly in the Caucasian population.

JOHN HOCKENBERRY: Dr. David Witt is a nationally known expert on cystic fibrosis.

DR. DAVID WITT: And there were issues about, can we educate people before they take the test adequately, so they know what they are getting into, what the limitations of testing are, can they be counseled effectively after they get their results if they are identified as carriers?

JOHN HOCKENBERRY: So far, more than 600 genetic mutations have been linked to cystic fibrosis. And, explains Dr. Witt, while the test for CF is highly accurate, if you know which mutation or set of mutations to look for, there is just no way to test for all of them. If you are a Caucasian from northern European ancestry, you have a one in 25 chance of carrying a single cystic fibrosis mutation.

DR. DAVID WITT: But when two carriers reproduce together, they have a 1 in 4 chance of producing an affected child

JOHN HOCKENBERRY: But what if you come from another ethnic background? If your ancestors came from southern Europe, the frequency of mutation is lower.

DR. DAVID WITT: When you get to the Hispanic population, the carrier frequency drops even more. And it drops even further in the African American population and even further in the Asian population, to the point of being probably negligible of being a real risk for the vast majority of people in the Asian population.

DR. MICHAEL KABACK: You can't expect everyone to be a geneticist, but certainly people should have a full understanding of what these diseases are; what these tests can tell them, how good are the tests, how accurate are they.

JOHN HOCKENBERRY: Dr. Michael Kaback is an expert on Tay-Sachs, which leaves it's victims in a chronic vegetative state until they die, usually by age four. Compare that to cystic fibrosis.

DR. MICHAEL KABACK: Cystic fibrosis can be, in some patients, nothing more than male sterility, with no symptoms other than that, not picked up until adult life. Cystic fibrosis can also be a very serious and lethal condition in early childhood. And there's all sorts of variation in between.

JOHN HOCKENBERRY: And, that's the problem. And for prospective parents that's the dilemma. Most genetic tests can't tell you the severity of the condition, only that it exists. And in the vast majority of conditions we can test for, medical science has yet to find a cure. Remember Judy? When we last heard from her, she had learned from her genetic counselor that she faced an awful choice. Here's Kathy McAnally.

KATHY McANALLY: When Judy found out that the child she carried had a chromosomal abnormality, that might or might not cause mental retardation and other health problems, her emotions went into freefall.

JUDY: Initially I thought that God was challenging me with this decision. That I've been given this challenge to live up to, to be strong; to take care of this child, no matter what. And then I looked at it the other way, and thought, you know what, we're living in a time where this information is available.

KATHY McANALLY: Judy says her first impulse was to blame the messenger. If she'd not been tested, Judy would have continued her first pregnancy, blissfully unaware that her child might be born with serious abnormalities. Barbara Katz Rothman is a Professor of Sociology at the City University Of New York's Baruch College.

BARBARA KATZ ROTHMAN: When somebody says, aw, let's just do the testing, nobody says, do you understand that we could find a condition that you would wish with all your heart that we had never told you about?

KATHY McANALLY: Professor Katz Rothman has written extensively about the psychological implications of prenatal genetic testing.

BARBARA KATZ ROTHMAN: Something interesting happened to pregnancy itself. A woman saying I am pregnant, publicly acknowledging that. By introducing testing that takes place late in the pregnancy, after weeks and weeks and weeks have gone by during which you're not drinking coffee and not drinking beer, and watching your diet—being

pregnant. So you have to go through all that time being in the social role of a pregnant woman, and saying, but I may not be having a baby. I may be having an abortion for a genetic defect. I came to think of it as a tentative pregnancy. You were pregnant but you could not yet commit to the pregnancy, it was a tentative pregnancy.

JUDY: I did get pregnant and I wanted to have that child, and yet after I lost... I decided to terminate that pregnancy, I felt there was no continuing my life without having another child. So I waited till what would have been the due date of my last child and had a ritual that we made up, friends and a rabbi, and after that, my depression lifted after her due date. And then I decided to try to have another baby. And I did. And I really felt this child was really meant to be born. The one I have.

[baby playing and cooing]

KATHY McANALLY: A woman with a wanted pregnancy faces one key question when she chooses to have prenatal genetic tests. What is more important: the quality of a child's life? Or the sanctity of that life? The answer depends on who you ask.

[Music, gospel-sounding, followed by sounds of baby and adults playing]

KATHY McANALLY: I'm on the living room floor at Lula Bizell's home in Opelika, Alabama. Tevin, a feisty toddler, is dancing and grabbing at my microphone. He's gonna be a preacher, says his proud mom. Mrs. Bizell has several other children, all born healthy. When she became pregnant with Tevin, her doctor recommended amniocentesis, since Lula was in her early 40s. She declined the test.

LULA BIZELL: Well, I really didn't trust it. You know, I had all my other kids and I was fine, so I felt that God blessed me with four, well three, and then he blessed me with this little boy, and I felt well, I believe in God, I felt, he's going to be alright.

KATHY McANALLY: And he is. Tevin is a healthy, happy little boy.

DR. SANDRA HEWLETT: When you offer a lady 35 years old and above amniocentesis for Down's, many of them turn it down. Because the average lady in the community does not believe in abortion, therapeutic or non-therapeutic.

KATHY McANALLY: Dr. Sandra Hewlett runs West Alabama Health Services, a network of clinics that serves a rural and generally poor population.

DR. SANDRA HEWLETT: This is the bible belt. It's not considered the thing to do. For any reason, health reason or the baby. If the baby's grossly deformed they believe that the baby is still a living human being, and if it lives to be born there's a reason for it to be that way.

KATHY McANALLY: Dr. Hewlett has been practicing here in Greene County, Alabama for nineteen years. I asked her how many of her patients had had an abortion after receiving bad news following a prenatal genetic test?

DR. SANDRA HEWLETT: I haven't had any.

DR. RUGAR STAGGERS: I've had one in my practice. One.

KATHY McANALLY: Dr. Ruger Staggers has been practicing in this area for 38 years.

DR. RUGAR STAGGERS: The idea of an abortion just does not sit well with them, and they therefore do not want to go through the trouble to go to Birmingham, to have the trouble to have the amniocentesis.

[Tour of University of Alabama Genetic Testing Center]

PAULA COSPER: We serve a very large population base, most of Alabama.

KATHY McANALLY: Dr. Paula Cosper directs the Medical Genetics Program at the University of Alabama in Birmingham. About 3,000 patients are seen each year, and the lab does over 2,000 prenatal chromosomal screenings annually. According to Dr. Cosper, very few rural Alabamans use the lab's services.

PAULA COSPER: I have spoken with some of the physicians in some of the more rural areas, and they tell me they tell their patients what's available. I think as long as they understand what's available, and what their risks are, if they choose not to have the testing, you know, that is their decision.

JOHN HOCKENBERRY: Choice: the right to make one's own decision about whether or not to test for recognizable genetic imperfections, is central to contemporary medical practice. *[old-time piano music]* But 100 years ago, those who concerned themselves with such matters were primarily focused on the goal of breeding better people. This foray into selective reproductive policies was named eugenics, and it became very popular in Great Britain and the United States. In the 1920s, a eugenics records office was created. Ruth Hubbard is Professor Emeritus of Biology at Harvard University.

RUTH HUBBARD: The job of the eugenics records office was to send out young ladies, trained at Radcliffe and Wellesley and places like that, to do interviews with poor families in order to develop a registry of debilities. And in this country as well as in Britain, such things as alcoholism and poverty and crime were all put together with hereditary deafness and hereditary blindness and all these various traits that clearly were bad for society.

JOHN HOCKENBERRY: Ruth Hubbard's interest in the American roots of the eugenics movement is fueled by personal history. Hubbard is Jewish, she was able to get out of Austria before it was too late. The Nazis invaded her homeland at the beginning of World War II.

[Snippet of Nazi music from the times]

TROY DUSTER: I think Americans need to hear this.

JOHN HOCKENBERRY: Sociologist Troy Duster directs the Institute for the Study of Social Change at the University of California at Berkeley.

TROY DUSTER: They tend not to know that the Germans and the Third Reich learned about sterilization in California.

BARBARA KATZ ROTHMAN: So the idea that there are some genetic weaklings that shouldn't reproduce and inflict their bad genes on the rest of the world is a very consistent notion.

JOHN HOCKENBERRY: Sociologist Barbara Katz Rothman.

BARBARA KATZ ROTHMAN: It got medicalized, it got shifted around. The word eugenics got dropped, but the concept that some people have bad genes and those genes should be stopped in their tracks and filtered out of our precious pool – that idea has been a consistent theme in the United States straight through the entire century.

JOHN HOCKENBERRY: Professor Troy Duster fears that prenatal genetic testing today is something of a back-door approach to new eugenic practices.

TROY DUSTER: Now the closer I got to it, the more I saw the technology was going to be used in ways... in respect to prenatal testing and diagnostics, and that some people will be screened more than others. That some will be thought more normal than others.

[*music*]

JOHN HOCKENBERRY: Who is normal and who's not? Producer Kathy McAnally picks up the story.

KATHY McANALLY: Ruth Ricker is a federal employee, a mother, a political activist. She is also not of normal height. Ruth Ricker is a person with achondroplasia. She is a dwarf.

RUTH RICKER: Achondroplasia is the most common type of dwarfness. The limbs are shorter than average in relation to the length of the body, so when I'm sitting, I'm sitting at about the same height as the average sized person but my arms and legs are shorter. And head size is usually average or slightly greater.

KATHY McANALLY: Achondroplasia is a dominant genetic condition. Dr. Claire Francomano is clinical director of the National Human Genome Institute in Bethesda, Maryland.

CLAIRE FRANCOMANO: Two people who have achondroplasia have a 25% chance of having a child who inherits the achondroplasia mutation from both the mother and father.

KATHY McANALLY: This is what's called getting a double dose, and it is fatal. A genetic test for the condition became available in 1994. Ruth Ricker was then the president of the organization Little People of America.

RUTH RICKER: Many of our families have two dwarf parents, and because achondroplasia is the most common type of dwarfism, oftentimes both parents have achondroplasia. We all know friends who have had children and did not live.

KATHY McANALLY: Many members of the short-statured community celebrated the availability of a prenatal genetic test for achondroplasia. But not all. Dr. Claire Francomano.

CLAIRE FRANCOMANO: It is not entirely lacking in controversy, because there are people among the short-statured community who feel that the use of prenatal testing has the potential to eliminate short-statured persons. There is quite a lot of emotion around this issue, and I think as a professional community we really need to be very sensitive to both the positive and the negative implications of this technology.

KATHY McANALLY: Ruth Ricker wanted a child. She chose to adopt a little boy with achondroplasia, rather than have a biological baby. She didn't have to decide whether or not to be tested. Choice is a key issue here. Geneticists and ethicists all agree that what separates modern genetic theory and practice from the bad old days of eugenics is that the individual retains the right to decide what to do. But according Professor Lori B. Andrews, that theory doesn't always translate into practice.

LORI B. ANDREWS: There are some very interesting studies now that suggest that if a woman doesn't undergo prenatal screening and has a child with a disorder that could have been diagnosed, the woman is blamed for that. I think genetic testing turns reproduction into production.

JOHN HOCKENBERRY: But exactly what sort of pressures are there on women to produce a better baby?

DR. GEORGE CUNNINGHAM: Now, if you ask questions about the public and even the legislature, there's a high acceptance of the woman's right to terminate a severely handicapped, defective fetus.

JOHN HOCKENBERRY: That's Dr. George Cunningham, Chief of the Genetic Disease Branch of the California Department of Health Services.

DR. GEORGE CUNNINGHAM: The lifetime cost of Down's Syndrome is about half a million dollars. The lifetime cost of spina bifida is something like 250 thousand dollars. These are expensive disorders to take care of and they drain resources from the family, so there's a considerable impact on the society.

JOHN HOCKENBERRY: But who couldn't be called a drain on society with the right economic analysis? In fact there are many families with profound and positive stories of how a child with a disability contributed to their lives in unexpected ways. Some parents say it causes pain to think that the prevailing view in society is that they would've been better off had their child not been born. Dr. Dorothy Wertz, senior scientist at the Shriver Center for Mental Retardation in Massachusetts, says that each family has to make its own decision as to what is serious and what they can live with.

DR. DOROTHY WERTZ: The only thing you can say is, frankly, that each family ought to have the fullest opportunity to see what life with a particular disability is like.

JOHN HOCKENBERRY: Dr. Wertz says she understands the fears of people with disabilities, that society will place less emphasis on research into treatments for certain conditions. But Dorothy Wertz also says that the technological advances in prenatal testing have gone hand in hand with improved services and civil rights for people with disabilities. And...

DR. DOROTHY WERTZ: Furthermore, you're never going to have disabilities disappear. Because most disabilities found at birth are not genetic. They are nothing you could find prenatally. Most disabilities at birth come from environmental toxins, lack of oxygen at the birth, some disease that the mother had while she was pregnant. So we will have plenty of people with disabilities.

JOHN HOCKENBERRY: In fact, prenatal genetic testing has reduced genetic disease in this country by only 5%. Still, researchers around the world will continue to hunt for the genetic mutations that cause a multitude of diseases, and develop tests to make a prenatal diagnosis. Today, new reproductive technologies such as in vitro fertilization allow a couple that carries the mutation for Tay-Sachs disease, for example, to choose embryos free of the disease for implantation. Dr. Michael Kaback.

DR. MICHAEL KABACK: They may produce five or six or ten embryos in the culture dish in the laboratory outside of the body. And then they can take a single cell from maybe a four or eight-cell embryo in the dish and determine whether that embryo is the one with Tay-Sachs or the one without. And then selectively only put back embryos into the mother's uterus that are free of Tay-Sachs.

JOHN HOCKENBERRY: Of course, this sort of technology doesn't come cheap. Professor Lori B. Andrews.

LORI B. ANDREWS: Certainly pre-implantation testing screening is only going to be available to the most wealthy. In addition to being able to screen embryos, we might move into an era where you can do genetic manipulation of embryos in petri dishes; and a survey by the March of Dimes found out that at least half the potential parents interviewed said they would be willing to genetically engineer their embryo to enhance its mental capacity, or to enhance their physical capacity.

JOHN HOCKENBERRY: Dr. Lee Silver, a Professor of Genetics at Princeton University, sees a future full of ethical dilemmas.

LEE SILVER: I don't see any problem with parents giving a gene to their child that will make their child resistant to disease. I don't see any problem with parents choosing an embryo that doesn't have cystic fibrosis or putting a gene into an embryo that protects that child from getting AIDS or heart disease or diabetes or obesity. What I do see, though, is the real ethical dilemma that's going to confront us in the future – is that this technology is very expensive. People with money are going to be able to give these genetic enhancements to their children, and people without money are not going to be able to afford it.

JOHN HOCKENBERRY: And so the moral of this story might be that a little knowledge is a dangerous thing. Unsettling for parents, ethically dangerous for society. Society has never been able to resist new knowledge, even when it comes with the kinds of dilemmas we've seen in this program. Prospective parents as well, from now on, will always have tough questions. Genetic testing makes parenthood an eerie roll of the dice. Parents who don't want to know their child's genetic future may have to live with a kind of high-tech intolerance over a disability that might have been prevented. There's no question that science will give us more prenatal tests, and, eventually, even the capability to treat fetuses, possibly with gene therapy. We can only hope that society will provide these new opportunities fairly... and that we take care to do no harm. I'm John Hockenberry

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Today's program, Prenatal Genetic Testing: Do You Really Want to Know Your Baby's Future? was produced by Kathy McAnally. The editor was Catherine Stifter and the engineer was Ed Herrmann, with feature production by Jon Kalish. This program is dedicated to the memory of Kathy McAnally's mother, Betty McAnally.

The DNA Files Executive Producer is Bari Scott. The Project Director is Jude Thilman.

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[music]

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