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Benjamin J. Shors

The University of Montana

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NOTES FROM THE ULTRASOUND:
A Survey of Prenatal Testing in Montana

by

Benjamin J. Shors
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Chairperson

Dean, Graduate School

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To my parents, for whom any list of what they've given me would come up far short; to faculty of The University of Montana School of Journalism for their guidance and trust; to Drs. John Johnson and Mary Haag of Shodair Children’s Hospital in Helena for their suggestions and dedication to the education of Montana parents.
Notes from the Ultrasound: A Survey of Prenatal Testing in Montana

Director: Clemens P. Work

This series culminates two years of graduate work at the University of Montana School of Journalism with an in-depth look at prenatal care in Montana.

The stories examine the social, ethical and pragmatic considerations of prenatal testing in an attempt to inform a wide audience of recent advances.

Several stories detail current services for expectant parents, explain basic genetic terms and examine current regulations for testing. These stories are important to parents, but in the rapidly morphing field of prenatal care, will quickly become dated.

Other stories give personal accounts from parents. These are stories of how parents make decisions. They are individual records of larger trends shaping the world's population.

Lastly, this series is an attempt to reconcile two parts of me: one which desires change and one which desires a continuation of the past.

It is, I suppose, two parts of us all.
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Two weeks waiting

Here's what amniocentesis will do for you: It will tell you if your child has a neural tube defect. It will tell you the sex of your child. It will tell you if your child will have Down syndrome.

It will not tell you what to do with those results.

A 1998 article in Genetics in Medicine charted amniocentesis procedures from 1972 to 1994. In the case of those fetuses that tested positive for Down syndrome, 86 percent of the parents chose to abort. Similar studies — and estimates by experts in prenatal care — bear out similar findings.

But as some ethicists are quick to point out, amniocentesis is not a cure for Down. It does not save lives. Despite the test, children with Down syndrome will continue to be born. It's just that, according to estimates, that event has become increasingly rare.

This is not a story about abortion. This is a story about a test that is shaping society. It is a story about a family. It runs from the spring of 1999 to the spring of 2000.
In the tungsten half-light of an exam room, the father stares from the darkness at the circle of light that falls on his wife’s round, full belly.

A doctor prepares to stick a three and one half inch needle into the stomach of Kristin Knudsen, 39, and withdraw 23 cubic centimeters of amniotic fluid. The fluid will be drawn into two vials, one to test for spinal cord defects in the developing fetus and one to examine the fetus’ karyotype, its genetic makeup.

“My daughter wants it to be a little girl,” Knudsen says as she lies on the exam table. On the screen to her right, images from her sonogram flash. The sonogram shows where the fetus lies and where the needle can be safely inserted. The child is lying on its back. Now it moves to its side. Its tiny arms swim in the bath of fluid. Its legs kick and flex. Its muscles generate surprising force. The pregnancy is in its sixteenth week.

Knudsen has already had two other sonograms.

“They said it looked ‘structurally sound,’” she says of the first one. “I said, ‘What are we building here, a garage or a baby?’”

The image is bouncing and twisting. The pixels on the screen somehow translate to life in the mother’s womb. The father leans into the circle of light, squinting at the screen.

“It looks like a boy,” says the doctor, though he can’t be sure.

The parents smile.

“He looks just like you,” Kristin says to her husband, Rowdy.
The sonogram has shown the doctor where the needle can be inserted. It takes only a few seconds.

Minutes later, the Knudsens are outside. They have a three-hour drive from Missoula to their home in Columbia Falls. Such is health care in Montana. It will be another 10 to 14 days before they know the results of the test.

It’s April in Montana and it hardly even seems worth mentioning that the snow is beginning to fall.

On average, one in 200 amniocentesis results in a miscarriage. The risk, experts agree, varies with the physician's experience. The more experience, the less risk.

The physician inserts a needle, which can be as long as 6 inches, into the woman’s abdomen, and withdraws the amniotic fluid surrounding the fetus.

The cells of the amniotic fluid have the same genetic makeup as the fetus. These cells can be tested for extra chromosomes or other genetic defects.

The amniotic fluid itself consists primarily of fetal urine and provides cushioning for the developing child. The needle removes two tablespoons of fluid, which the fetus replaces in a matter of hours. At 16 weeks, nearly a liter of the fluid surrounds the child.

The Knudsens head north on Highway 93, home to wait for their results. The sample takes a Greyhound bus east (but we’ll get to that later).
Highway 93, the Knudsens’ route, is not just another road. By many accounts, it is one of the most dangerous roads in the western United States.

Several years ago, National Geographic featured a story on the road. It ended with a plaintive plea: “I drive Highway 93. Pray for me.”


The mountains here are magnificent, a wall of black rock streaked by arteries of snow and chutes. The rocks were laid down about a billion years ago, when an inland lake covered the area. The mountains are here because a plate from the Pacific Ocean crashed into the North American landmass some 80 million years ago. It pushed up mountains running from Alaska to Mexico. The chutes stream down the mountains because of the avalanches. And the avalanches, well, they come with the snow.

Columbia Falls sits at the base of these mountains.

It is not always an easy place to live.

“Last winter, there wasn’t hardly any snow here at all,” Rowdy says.

But the winter before that, the winter of 1996, the snow fell long and the snow fell hard. It started slow enough, grand white shavings as the summer’s light shrunk away,
but it just never seemed to stop. It covered roads, it filled pine trees, it reached to the
second floors of houses and cabins. Land in Kalispell and Columbia Falls suddenly went
up for sale. People left for warmer climates.

The Knudsens stayed.

The sample is loaded into the luggage compartment of a Greyhound bus and
heads east 200 miles to Helena and Shodair Children’s Hospital, the only genetics lab in
the state.

If the outside temperature gets too cold or too warm, the driver carries the
package underneath his seat. The sample needs to be kept at room temperature.

When the sample reaches Shodair, the cells are spun at 900 r.p.m. until the fetal
cells separate from the rest of the solution. The cells are placed on a slide in a petri dish
with growth media. The cells divide and divide and divide.

After a week, the cells are ready to harvest. A chemical freezes the fetal cells in
midpoint as they’re dividing. Acetic acid ruptures the cell membrane, spilling the
chromosomes, which can then be stained, counted and examined.

Rowdy Knudsen has hands that build, an important tool for a man working in the
construction business. Those hands move from his face to his mouth to his head,
readjusting his red ball cap, scratching his face, brushing his cheek as his wife discusses
her pregnancy.
It’s 6:30 on a Sunday night at the kitchen table and the Kristin and Rowdy are halfway through their wait for the test results.

Kristin has just finished her shift at Costco where she splits her workday between manning a checkout line and selling tires in the shop. Rowdy has come in after putting siding on the house all day. Their daughter Shiloh, dressed in bright pink shorts with bright red lipstick, floats in and out of the conversation. She is 8.

“She worries about being fat and being pretty,” her father says.

His daughter cried for a few hours when her parents broke the news that, according to the ultrasound, her newest sibling was most likely a boy.

“She was not accepting the fact that it was anything but a girl,” says Kristin. “She was disappointed, but she gets to be the big sister now.”

Shiloh is something of an only child, though Rowdy’s two sons from his first marriage visit regularly. At 8, it can be hard to realize that your parents don’t have control of everything. Some things are out of reach.

“At that age, you just think that everything’s perfect,” Kristin says.

By the age of 39, reality is a little harsher. For women in their 20s, according to the National Association of Down syndrome, the abnormality occurs in one in 1,250 children. When a woman reaches the age of 35, the risk of having a child with Down syndrome is one in 350. By age 40, the risk is one in 100.

But those are only statistics, abstract numbers.
For Kristin, the test itself went well, much to her relief.

"I’ll tell you what, I was more nervous about taking the test than I am about the results," she says.

Fortunately for the Knudsens, Kristin’s insurance covers the test, which otherwise would have cost them $675. But Kristin says that doesn’t matter.

"Sometimes you wonder about how much things are going to cost," she says. "But sometimes you just don’t. When your kids are concerned, you don’t."

Whatever the results of the test, the Knudsens say they’ll keep the child.

"We probably wouldn’t consider abortion," Rowdy says. "Kids have just as much a right to live as anyone else."

He looks out the window; the view from their two-story home is not of mountains so much as it is of neighbors and trucks and backyard swingsets.

"The way I see it," Kristin says, "if there’s a problem with the baby, that was decided a long time ago, by someone with a lot more power than me. The Lord had already decided. To me, it’s the way it’s supposed to be and you’ll deal with it. I believe in God and that he’s not going to give you more than you can handle."

Chromosomes contain genetic information from both the mother and father.

*There are normally 23 pairs of chromosomes in a cell. In Down syndrome, or Trisomy-21, the 21st pair has a third chromosome. In other abnormalities, there may be only pieces of a chromosome.*
Through the lens of a microscope, technicians photograph the stained chromosomes, now magnified 1,000 times. They transfer the photo image to a nearby computer, where the chromosomes are arranged by size and distinguishing stripes. A lab technician counts and measures them. Several samples are examined to decrease the chance of error.

After the test, the results are returned to the doctor who then informs the parents.

A week later, the news is good.

“It’s a great big relief,” Kristin says. “I mean no matter what happened, we were going to keep the baby and love him, but I’m very happy that everything’s normal.”

It’s the first day in May and even the mountains are readying for summer. The snow is gone or in patches at best. The runoff has raised the rivers; they will climb even higher in the coming days. Children and teenagers and college students are surviving the last weeks of classrooms and lectures.

And the Knudsens have their results.

The child, the test says, has the chromosomes of a normal, healthy baby boy - that’s all it can determine. He’s scheduled to arrive in late September.

“I was a little nervous,” Kristin concedes. “But it’s good to know.”

Even Shiloh is warming up to the prospect of a baby brother. Maybe by knowing a few months in advance, Shiloh will be better prepared for the coming adjustment, the new baby.
The amniocentesis did more than just identify the sex of the child. It provided peace of mind. It gave the family a chance to prepare. It gave the family, no matter their final decision, a choice.

“I think [the test] is a real good thing,” Kristin says. “Every mom should have a chance to have it.”

Mother and son are at nineteen weeks. Only a stretch of welcoming summer separates them, if anything does.

“He’s getting bigger each day,” Kristin says. “I can feel him kicking to get out.”

Amniocentesis and other prenatal tools have spread around the world. Responses to the test are as varied as the countries themselves.

In Ireland, where children with Down syndrome are called “the people of God,” abortion is illegal, and amniocentesis is not widely used.

In India, a drop in the country’s female population was attributed in part to the growing practice of aborting female fetuses after amniocentesis indicated their sex. In 1994, the government passed a law banning the practice under the Prenatal Diagnostic Techniques Act. UNICEF workers in India report that the law has gone largely unenforced.

In most African countries, geneticists are scattered and rare. Poverty makes survival the first priority.
As her child's due date nears, Kristin and Rowdy find themselves at the doctor's office twice a week. Doctors worry about Kristin's elevated blood pressure and the child's movement. Kristin undergoes fetal movement tests - an elastic belt wrapped around her belly, a fetal monitor tracking the child's movement - every week for the last two months of her pregnancy.

On the day she enters the hospital in late September, Rowdy is working two hours away, in tiny Essex, Mont. She calls. He rushes home to take her to the hospital.

The contractions are strong and spaced just minutes apart, but there's no delivery. Doctors and nurses continually check on her, but nothing seems to be happening.

Finally, on the fetal heart monitor, Kristin and Rowdy watch as their son's heart rate drops from 100 to 40. The doctor gives it a moment to recover. When it continues to hover at 40, he quickly prepares for a caesarian section.

"He started shouting 'stat this and stat that,'" Kristin remembers. "Rowdy and I knew what that meant ... we watch E.R.

"Everyone kept saying relax, but of course you can't relax in a situation like that."

Gabriel Knudsen - 6 pounds, 10 oz. - arrives just before 7 p.m. and the doctor rushes him from the room to have his lungs suctioned. Ten minutes later - though "it seemed like forever" - Rowdy cradles his healthy son in his arms.

"All the tests we had and everything, they were great," Rowdy says. "But you just can't know everything that's going to happen."
In a few years, maybe five to 10, health expects say an expectant mother may walk into a doctor's office, and have her blood drawn - no needles in the belly, no risk to the child. Amniocentesis, health experts agree, will be a dinosaur.

The fetal cells that circulate in the mother's blood will be extracted and examined, the information available to parents and doctors with no more risk than donating blood.

Arthur Caplan, director for the Center for Bioethics at the University of Pennsylvania, goes even further.

In this century, Caplan predicts, parents will use genetic engineering to give their children a jump on other babies, and "embryos will be tested for genetic diseases and subsequently grown in artificial environments to maximize their health and potential."

A revolution in prenatal care is coming, the experts agree.

A revolution.

All of a sudden, it seems, it's April again, and Rowdy and Kristin rest at the kitchen table.

A room away, 7-month-old Gabriel is sleeping off his dinner. Betty, the shaggy black family dog, climbs on the picnic table in the yard to regard through the window the remains of dinner – grilled chicken breasts, scalloped potatoes, cooked carrots, dinner rolls.

The house is relatively silent, and Kristin rests her hand on the knee of her husband’s black jeans.
Before “this” all began, the Knudsen family once took a vacation to Mexico, and they remember it well, they retell it well:

When you ask for a drink at poolside in Matzatlan, Kristin says, the waiter brings you two. And there’s sleeves of fresh donuts for nickels and dimes at local bakeries. And the scuba diving? The warm saltwater buoys you up so you don’t even have to paddle - you can just float.

We’ll go back, they promise each other. Perhaps next year, they agree.

From the other room, young Gabriel begins to stir from his nap.

It’s early evening in Columbia Falls, and there’s light that remains from the day, hanging over the valley and stretching into the house.

Kristin rises wordlessly from the table and walks through the archway to the clean, well-lit living room to retrieve her waking son.
What’s available?

Prenatal testing can leave both parents and doctors with more questions than answers. A guide to what parents need to know about current tests.
When a screening test indicated that the child Mary Ann McGowan carried had an increased risk of having Down syndrome, the 32-year-old Missoula woman decided she already had enough information, and opted to forego further testing that could have told her definitively her child's condition.

"I was okay not knowing until I delivered my child," said McGowan, whose son Daniel, born last spring, has Down syndrome. "Some people need more and more information, but I think it would have caused too much worry. It would have put a cloud over the pregnancy."

Her choice - opting for less information even as medicine provides parents with more and more knowledge about the health of their fetus - may seem unusual.

Shodair has screened 30,000 expectant mothers since 1990. In a state of nearly 150,000 square miles, nine out of 10 prenatal and genetic tests are routed through the clinic at the eastern edge of Helena.

"Our philosophy is to give the information to the parents and then let them decide," said Dr. John Johnson, the clinic's director.

But a 1998 study from the clinic itself indicates that many Montana women whose pregnancies are at risk don't always want prenatal tests and the answers they provide. After decades of advances in prenatal screening and testing, doctors and ethicists are often unsure which tests should be offered to which patients – and many patients are unsure if they want them.
Following a screening test that indicates their fetus may have a chromosomal abnormality, the Shodair study showed only 50 percent of Montana women chose to have an amniocentesis test that could tell them definitively the condition of the fetus.

Doctors, too, are sometimes unsure what the tests can provide patients. And there's a growing apprehension that some patients may blame doctors if their baby is born with a disability.

‘Wrongful birth’ lawsuits – including cases where parents sued their doctor for failing to detect the child’s disability in utero – have sprung up across the United States, though none have been filed in Montana.

Johnson said many patients and some physicians are confused about how to interpret the results of a screen test and a diagnostic test.

A screening test - such as a maternal serum triple screen - indicates only that the baby is at an increased risk. A diagnostic test - which gives the parents a definitive answer - is generally recommended for parents wanting to know the health of the developing child but may put the fetus at a slightly higher risk of miscarriage.

The knowledge that their child will be disabled forces parents to decide whether to carry the child to birth or terminate the pregnancy.

"[Parents] should have a little counseling before the tests so they understand what they’re getting into," Johnson said.

Herewith, a guide to the risks and considerations of current prenatal tests.
Ultrasound

At dozens of Internet sites, expectant mothers have gotten a jump-start on their baby’s photo album.

The ultrasound photos are grainy black-and-whites, and most have been outlined or diagrammed to highlight distinguishing features. Here’s a foot, a nose, a hand.

The photos that haven’t been highlighted resemble a faded Rorschach ink blot test, and its meaning may be just as mystifying to parents.

“It’s not exactly easy to tell what you are looking at when they are so little,” one mother wrote.

The message of the pages is clear and intimate: These pixels prove there’s a child within the mother.

“An ultrasound does two things,” Johnson said of the screening test. “It immediately establishes a relationship between the mother and the child, and, if there’s a problem, the parents are looking at it at the same time as the doctor.”

Ultrasound creates an image using sound waves and is generally accepted as being very safe - though there have been no long-term studies on its effects.

At the Fetal Diagnostic Center in Billings, parents have the chance to view computer-generated images of their fetus using a 3-D ultrasound, the only machine in Montana and one of only a handful in the United States.
"We can get a better idea of where the defect is," said Leslie Brown, a genetic counselor at the center. "The parents really love the 3-D ultrasound. It makes them feel more reassured [about the child’s health]."

The 3-D ultrasound takes several pictures of the fetus and then forms one image. The procedure is the same as a conventional ultrasound - the only difference is the computer provides a third angle.

"It’s a very different way of looking at the baby," Brown said. "When you’re looking at the pictures, it looks very real to the parents."

Adding a third angle gives depth and shading, resulting in a picture more recognizable to parents.

The procedure may prove most effective at screening for Down syndrome by measuring the thickness of the skin at the babies' neck. An increased thickness means a higher risk of the abnormality.

By measuring the thickening, doctors can decide whether to recommend a diagnostic test - such as amniocentesis or chorionic villus sampling - for the fetus. The ultrasound can be done by the 10th week, giving parents more time to decide whether to undergo further testing and prepare for the results.

But ultrasound images are often grainy, making it hard for the doctor and parents to distinguish the fetus’ sex or physical abnormalities.
"The skill of the operator makes a big difference in detecting problems," Johnson said.

The new technology raises ethical issues, as parents must decide what to do when the test indicates an abnormality. Some disorders, such as webbed fingers or cleft palate, can be corrected by surgery and aren't life-threatening.

"The better the ultrasound is, the more likely a person is to know about cleft palate," said Amy Mackin, hotline manager for the Cleft Palate Association in Chapel Hill, N.C.

Mackin said although some callers have discussed abortion after an ultrasound showed a cleft lip, she hadn't noticed any trends in the United States.

"In places where abortion laws are different or where standards of acceptance are different than they are here, the situation may be different," Mackin said.

At the Hawaii Birth Defects Registry, administrator Ruth Merz oversees one of the world's most comprehensive programs for tracking birth defects.

"Hawaii is a very liberal state," Merz said. "We tend to have a higher incidence of terminated pregnancies for abnormal fetuses than most states do."

From 1986 through 1998, the registry recorded 535 prenatal cases of oral cleft. Of those, two were aborted because of cleft lip or palate.

"[The decision to terminate] can depend a lot on ethnicity," Merz said. "Some ethnicities seem to really want a perfect baby and they have no problem terminating a
pregnancy and trying again. Other ethnicities, depending on their personal beliefs, will bring the baby to term and then have it surgically corrected."

A study of Israeli women, as reported last spring in the *Cleft Palate-Craniofacial Journal*, found that 23 of 24 women terminated their pregnancy after learning in the 15th week that their children had cleft lip.

The timing of the ultrasound, as well as culture, influences the outcome of the pregnancy.

In a similar San Diego study, two of eight women had an abortion. However, the San Diego women didn’t learn the results until the 24th week – after which many doctors won’t perform an abortion.

**Maternal serum triple screen**

Of the 12,000 pregnancies in Montana each year, Shodair screens about 25 percent – a rate less than half that of some other states, Johnson said. In California, for example, screening is offered by mandate and about 60 percent accept.

The maternal serum triple screen, a medically simple blood test, measures the levels of three markers in the amniotic fluid. Taken together, the levels can indicate but not diagnose spina bifida and Down syndrome, as well as abdominal wall defects and other disorders.
“The problem that some people have with screening, is that with the percent that are positive, it raises anxiety,” Johnson said. “Both doctors and patients have to understand it’s a screening test – don’t panic.”

Johnson said with Down syndrome, a result is “positive” if the risk is greater than 1 in 300 – which is the predicted risk for a 35-year-old woman. He said 95 percent of the screen positive tests turn out to be normal.

The triple screen, which costs $80, detects Down syndrome in 60 to 70 percent of women under 35, and nearly 90 percent in woman over 35.

The procedure is typically done in the 15th or 16th week of the pregnancy – allowing time for a definitive result from amniocentesis or chorionic villus sampling.

**Chorionic Villus Sampling**

Although chorionic villus sampling (CVS) diagnoses fetuses with Down syndrome earlier than amniocentesis, it is twice as likely to result in a miscarriage.

CVS, which can be done between the 9th and 12th week of pregnancy, has also been linked to limb defects – though recent studies have proved inconclusive.

CVS requires a tube be inserted into the cervix to remove tissue from the chorionic villi, which later becomes the placenta.

The procedure only tests for chromosome abnormalities – amniocentesis also indicates neural tube and abdominal wall defects.
Amniocentesis

A powerful diagnostic test for decades, amniocentesis is widespread and accurate.

The test causes a miscarriage in 1 in 200 pregnancies, and is done between the 14th and 18th week of pregnancy – before the halfway mark of the pregnancy to allow parents time to decide how to handle a positive result.

A fine needle is inserted in the woman’s abdomen and withdraws amniotic fluid containing cells from the fetus. The cells are grown in the laboratory and studied for complications such as Down syndrome, spina bifida and abdominal wall defects.

The test is generally recommended for women over 35 – older mothers are at a significantly higher risk of having a child with Down syndrome – or where there is a family history of genetic disorders.

The test remains relatively unchanged since it was introduced 25 years ago.

“I think the thing that will make a huge difference – and it’s still years away from clinical practice – is testing maternal blood for fetal cells,” Johnson said.

The test would be as safe as drawing blood and could be conducted earlier in the pregnancy, allowing parents more time to consider whether to continue the pregnancy.

Several national studies have shown that close to 90 percent of women whose amniocentesis indicated abnormalities in the fetus chose to terminate the pregnancy. A
1998 study in *Genetics in Medicine* correlated a drop in the number of Down syndrome births to the advent of amniocentesis - which was introduced in the early '70s.

Johnson estimated a dozen of Montana’s 3,000 annual abortions resulted from prenatal testing. In 1997, the Helena clinic had 18 abnormal tests, not all of which led to termination of the pregnancy.

The procedure, which costs about $500, also identifies the sex of the child, allowing parents to abort a child because of its sex.

In India, the government banned in 1994 the growing practice of aborting female fetuses after a census showed skewed sex ratios.

In 1991, India had less than 93 women for every 100 men - a figure affected not only by new medical technology, but also by infanticide and neglect. Despite advances in women's rights, the ratio was lower than it was in 1901.

In the United States, England and France, the United Nations reports 105 women for every 100 men.

UNICEF officials in India said enforcement of the 1994 ban “has not been very extensive” in penalizing hospitals and medical institutions.

Johnson said he didn’t know if any of Shodair’s patients had used the test to selectively abort a fetus because of sex, and that patients aren’t required to give reasons for having an abortion.
Johnson said that since abortion is legal regardless of the child's sex, there is no ban on sex-selective abortion in the United States.
**Q and A**

To understand issues in genetic testing and prenatal care, you have to get a handle on the basics.
You’d be lucky to go a day without hearing about genes and the implications of research surrounding them. And if you’re about to have a child, you may notice it even more.


The reports may tempt you to eventually dust off that dog-eared biology text buried in the garage, but in the meantime, here’s a guide to the basics:

Q: What are genes?

Inside a cell, 23 pairs of chromosomes drift in the nucleus. Those 46 chromosomes contain coiled genes – the chemical messages of heredity – working subunits of DNA that each code for a specific product – usually a protein such as an enzyme.

There are some 140,000 genes in the human genome, but a misstep at one can have dramatic effects: deformities, disease and sometimes death. If a gene is mutated or deleted, a protein may not be made or may function improperly.

Each gene is made up of hundreds of thousands of chemical bases - represented by A, T, G and C. The order in which the bases occur, like a biochemical recipe, determines the gene’s function.

Q: Why are chromosomes paired?
Everyone inherits two sets of 23 chromosomes – one from each parent. They differ in length and width and the information they provide. Human cells contain two sex chromosomes – mothers can pass on only an X, while fathers pass on an X or a Y.

An X chromosome is longer than a Y chromosome – a feature that can be detected by researchers and used in pregnancies where parents want to choose the sex of their child.

Q: **What do genes do and how do they do it?**

Each cell (except red blood cells, which have no nucleus) contains all the body’s DNA information. But cells activate only the genes they need and suppress the others.

A gene in a bone cell may produce an important protein, but the same gene in a brain cell may be inactive most of the time.

Some genes function in development during pregnancy, and then shut down.

Q: **What is gene therapy?**

Gene therapy is used to treat disease by inserting genes into the body’s cells to replace or manipulate a gene that isn’t working correctly.

It’s a process that relatively easy to do at an early stage of development – such as a fertilized egg. The changes made at that point will be replicated billions of times and passed on to other cells in the body.
But by birth, any mistake in a gene has been passed on to all the body’s cells, making a genetic anomaly much more difficult to treat.

So scientists use viruses – such as a common cold virus – to invade the body’s cells and inject the DNA they carry.

Rather than carrying DNA that causes disease, the viruses have had their DNA replaced by the genes needed by the body. The new genes, researches hope, will take over for the nonfunctioning gene.

Q: Does it work?

Over the last 10 years, gene therapy has been tested on 4,000 people – with little success.

Originally hyped as a breakthrough in genetic diseases, gene therapy has failed to produce the results many hoped for, in large part because scientists have had an unexpectedly difficult time getting genes to the organs where they are most needed.

Until last fall, the trials seemed harmless. But in September, an 18-year-old man died at the University of Pennsylvania during a gene therapy trial designed simply to test the safety of the procedure.

The death has been a public-relations disaster for the field of gene therapy, in part because, unlike most deaths in clinical trials, the man was not terminally ill and was on
medication that controlled his disease. It was the experiment, not the disease, that killed him.

The Food and Drug Administration, which approves all gene therapy trials, suspended the Penn researchers.

But the hope that gene therapy will one day revolutionize health care remains.

In perhaps the most promising trial in gene therapy, researchers last March at Children's Hospital of Philadelphia announced that two patients with severe hemophilia had increased levels of an important clotting factor after being injected with a gene to help treat their bleeding disorder.

Q: How has this changed pregnancy?

By best guess, there are currently prenatal tests for over 200 genetic conditions, a far cry from a parent's option 25 years ago.

One goal is a simple maternal blood draw that would identify genetic disorders in the first weeks of pregnancy.

Testing is only part of the advance. Doctors hope by identifying genetic problems early, gene therapy or fetal surgery (done within the womb) will be able correct the problems in utero.

Q: What is the Human Genome Project?
In 1990, the Human Genome Project - a collection of hundreds of scientists and researchers around the world - was established to find the location of the 100,000 or so human genes and map the entire genome by the year 2005.

The human genome, which consists of 3 billion chemical bases, would fill 1,000 thousand-page phonebooks. Finding a gene that causes a genetic disease - such as Huntington's disease, cystic fibrosis or Duchenne muscular dystrophy - would be like finding a needle in a stack of haystacks.

To speed up the search, much of the work has focused on the development of new techniques and technologies, and continued advances in computer processing have accelerated work.

The project to map the genome is a large one - the tab to taxpayers is $3 billion - but it's not the only search of its kind.

At Celera Genomics in Maryland, Dr. Craig Venter is betting his company can decode the genome quicker than the government's project. The venture - with a privately funded budget of $200 million - has promised a complete map by 2001.

The competition has forced the Human Genome Project to pick up its pace, predicting a complete map by 2003. Both groups say they'll have a rough draft ready by this spring.

Q: How will the information help doctors and patients?
Even with the map in hand, the genes won't tell researchers how mutations lead to disease or how they can be treated.

There is no "normal" genome after all. Each person has a distinct genetic makeup, so researchers with the Human Genome Project are examining several different genomes.

Once researchers determine the sequence, they'll compare variations in different people and determine which differences are associated with disease - an incredibly complex process given that there may be 3 million differences in the sequences of two unrelated people.

Only a small portion of those differences cause disease. The rest determine absolutes like eye color or height, as well as less tangible features such as intelligence and talents.

Finding and understanding the function of genes can help scientists attack a disease at its root and tailor treatment to the individual. New drugs may then activate or suppress certain genes in a novel approach to treating both physical and mental ailments.

Doctors are already able to do some predictive testing - examining a person's genetic makeup in an attempt to determine the likelihood of developing a certain disease. Knowing the function of the body's individual genes will exponentially expand the number of diseases that can be diagnosed before symptoms appear.

Q: What is genetic screening?
While gene therapy exists only in clinical trials, genetic screening or predictive testing for diseases such as Tay-Sachs, cystic fibrosis and ALS (Lou Gehrig's disease) are widely available.

Genetic tests examine a person’s DNA for changes or anomalies that signal a risk of developing a disease.

Genetic tests deal in probabilities, not certainties. For example, women who have the BRCA1 breast cancer susceptibility gene have an 80 percent chance of developing breast cancer by the age of 65.

But inherited forms of cancer represent only about 5 or 10 percent of all cancers. The rest result from acquired mutations that develop during a person's life and can't be passed on to their children.

The tests are safe, involving a blood draw or occasionally samples of body fluids or tissues.

But the psychological risks of a test indicating a predisposition to cancer or disease can be devastating, particularly because the test may provide information not just about one person, but about an entire family.

Q: Is there anyone to help navigate me through genetic testing?

Genetic counselors help patients determine what tests are available, whether a patient should have a specific test, and what the results mean. They also work with the
families of children with disabilities to make sure unique developmental and nutritional needs are met.

"We're kind of the front-line for people who have any questions about genetics," said Katherine Berry, a genetic counselor at Shodair Children’s Hospital in Helena.

Berry and two other counselors from Shodair travel to one-day specialty clinic around the state, where parents can arrange appointments to discuss cleft palate, heart defects or developmental delays.

As testing expands, the demand for counselors grows.

Currently, there are only a handful of counselors in the state and approximately 1,000 nationwide.
The search for answers

In Eureka, Montana, five children of local teachers – plus the child of a high school student – have been born with birth defects in the last six years. No one knows why, and state health officials say no one’s likely to find out. What happens when prenatal testing misses problems in a pregnancy?
Last September, a few hours after the birth of her daughter Peyton, Rae Lynn Benson tried to feed the tiny newborn, only to watch the formula rush out through Peyton's nose.

Thinking her daughter's cleft cheek prevented her from feeding, Rae Lynn tried again. This time, Peyton turned blue.

Moments later doctors rushed the child off. By the next morning, they had diagnosed Peyton with tracheoesophageal fistula (TEF), a condition where the child's esophagus leads to a "blind pouch" rather than the stomach.

Benson, of Eureka, Mont., had a triple screen and three ultrasounds during her pregnancy, but her child's health problems went undetected during prenatal testing - despite decades of advance in screening and testing. Each year in Montana, 300 to 400 children are born with birth defects, only a fraction of which can be detected by prenatal testing.

"It would have been nice to have been more prepared when she was born," Benson said. "Instead, we were fumbling around, trying to figure out what had happened to our daughter."

At the Kalispell hospital, Benson said neither doctors nor nurses told her of any support groups in the state, provided counseling, or mentioned the need to register her daughter with the National Birth Defects Registry. Weeks later, she learned that Montana didn't have a birth defects registry - and no way to track problems like her daughter's.
“We kind of felt lost,” Benson said of the days after her Peyton’s birth, “like we were heading into the unknown.”

With an ever-growing focus on identifying birth defects through prenatal testing, the Eureka parents are raising some interesting questions about the care of those children testing misses.

**Problems in cluster investigations**

The Bensons, who both worked for the Eureka School District, were not the first family in Eureka to have a child with health problems. Five children of Eureka teachers – plus the child of a high school student - have been born with birth defects in the last six years.

Frustrated and frightened by the problems, the parents began looking for help. In the process, they’ve challenged the state’s ability to track and care for children with birth defects – the leading cause of infant mortality in Montana and the nation.

Dr. Michael Spence, state medical examiner, said he can’t tell if there’s a cluster of birth defects in the school or if the problems are just a statistical quirk. Spence said that in the vast majority of cluster investigations, investigators are unable to find an environmental cause.
In Eureka, Spence must determine first if the problems are in fact a cluster, which requires comparing Eureka births to birth defects in the surrounding area, a difficult task without a registry.

Spence, who was contacted by Eureka parents last fall, began the investigation by trying to track birth certificates at local hospitals.

Birth certificates have a listing of major birth defects, but the information is often incomplete and rarely analyzed, according to state and national health officials. The information from birth certificates is sent to the Office of Vital Statistics.

“A lot of this information comes in to the office,” Spence said, “but the majority of the people that it comes in to, that’s not what they’re looking for – they’re looking at making sure it’s accurate and electronically stored, and they put it away. Their job is archiving, not looking at the registry.”

Jackie Wynne, community liaison for the California Birth Defects Registry, said birth certificates record births within a strict legal timeline and aren’t designed to track problems.

“California did a study which showed that birth defects are only reported on the birth certificate 19 percent of the time,” Wynne said. “Even very obvious birth defects, like anencephaly and cleft lip and palate, were missed frequently. Of those that were recorded on the birth certificate, many were not actually a birth defect and others were misclassified.”
Unable to use the birth certificates – which were scattered and often incomplete –, Spence asked the Eureka parents to track birth defects in the Tobacco Valley by calling the parents of children under 10 years old.

The parents agreed but say they already feel overwhelmed caring for their children and under-trained to interview other area families.

"As parents, we feel like we already have had enough to deal with," said Benson, whose daughter's hospital bills are nearing $100,000 after five months. "Each of the children are requiring additional care so we're still dealing with the clinics, and we're dealing with all the medical bills and everything else, and it feels like this is just one more thing we have to deal with. I guess we incorrectly assumed that this was something the state would do."

'A failing grade'

In December, Montana received a failing grade from a major environmental-health organization for the way it tracks birth defects. But health officials with the state say that poor record is about to change.

Montana received a three-year federal grant last year to start a registry, and state health officials say a pilot birth defects registry will be tested this summer and should record all births for 2000.
"We had a lot of things to do behind the scenes to get this up and running," said Jan Baker, newborn screening program manager at Montana Special Health Services, "It just was never done before because there weren't the bodies to do it and there wasn't the money."

The program will track birth defects in four areas - neural tube defects, cleft lip and palate, congenital heart defects and congenital hypothyroidism. Baker said those areas were chosen because support services were already in place to educate parents.

Baker said Down syndrome and other common congenital abnormalities would be tracked but could not say in which category they would be classified.

After three years, Montana will have to apply for another federal grant or fund the project with its own money. The grant provides about $90,000 a year.

Baker said Montana's records were "very incomplete" and a "best guess."

"By collecting the information," Baker said, "if we have something that looks definitely different from the national average for a particular defect, then we can say, 'This doesn't look right, maybe somebody should look into it.'"

Baker said results in the first few years will likely be skewed until a significant number of births have been tracked. It could take years before workers can track trends in birth defects and possible causes.

Dr. John Johnson, director of Shodair Hospital's genetics clinic, said the results could be used to suggest screening tests in areas showing a high incidence of birth
defects, but the registry’s primary purpose was to ensure the diagnosis and care of children with birth defects.

"Right now," Johnson said, "we just don’t have any information on birth defects in Montana and their frequency."

Why is a registry needed?

In giving the state a failing grade for its tracking of birth defects, the Pew Environmental Health Commission said Montana’s lack of a birth defects registry was a major reason that increases and trends in birth defects can’t be explained.

"Without this information, public-health officials are working in the dark,"

Lowell Weicker, commission chairman and a former U.S. senator, said in a press release announcing the results. “We lack the key tool needed to identify emerging disease clusters and trends, making it tougher to tackle environmental threats that may cause sickness and death in our children.”

The study gave failing grades to one-third of states, Puerto Rico and Washington, D.C., for what the organization considered the lack of a registry. Only eight states received an “A.”

Montana’s registry most likely would not be used to identify clusters. Its primary purpose, officials say, will be to monitor rates and trends of birth defects, and to identify cases to be used in large case-control studies designed to identify causes of birth defects.
Dr. Spence, lead investigator in the Eureka case, said a registry might have alerted health officials earlier of the possible cluster.

**Birth certificates inadequate**

At the Office of Vital Statistics in Helena, research specialist Bruce Schwartz oversees the nearly 11,000 birth certificates that come into his office each year. The office also handles deaths, marriages, divorces, and induced abortions, as well as operating the Montana Central Tumor Registry, which tracks cases of cancer.

“We don't concentrate just on [birth defects],” Schwartz said. “We deal with all items from all certificates and reports. I don't have nearly enough time to concentrate on this.”

The bureau’s primary duty is making sure the certificates are correctly filled out, Schwartz said, not searching the data for trends. Without analyzing trends, state health officials may not notice possible clusters, such as Eureka.

But even closer analysis of available information may not help.

Birth certificates must be filed within 10 days of the birth, and Schwartz said most are completed in the first two or three days – before doctors are even able to identify some health problems.

“[Birth certificates] are comprehensive in the sense that a lot of different birth defects can be recorded, but many of them simply haven't been diagnosed by the time the
mother has been discharged,” Schwartz said. “We think things are probably under-reported.”

Schwartz said until the registry is ready, birth certificates are the state’s only way to track problems.

“All in all, I guess you could say we’re not quite satisfied with what we have, but I’m not sure what we would do to improve the system.”

An uncertain future

Chocolate cupcakes are piled high on a table at the back of Angela Price’s second-grade classroom in Eureka Elementary School. The class is preparing for a President’s Day celebration.

As her students work, Price—now seven months pregnant—sits at her desk and takes a drink of the water she brought from home. It’s one of the precautions the young teacher is taking. One of the many rumors in the school tied the birth defects to lead in the water. Others blame asbestos or mold or the school’s archaic heating system. Still others say it’s just bad luck.

Price’s room is a few feet away from Larry Benson’s. A few feet farther down the hallway is Korena Henry’s room. Henry’s son was born with aortic stenosis and is six-months removed from heart surgery.
"Almost every friend I have has a health problem with their kids," said Henry, an elementary teacher whose husband teaches at the high school. "We’re doing this not to try to scare the young teachers, but because if there is a problem, we need to find out."

Parents say the investigation has stalled, that there have been communication problems, that state officials are too busy with other investigations. The parents say they’re considering doing their own testing of the school.

Price’s child is due in April. So far, she’s avoided prenatal tests because she’s not sure what she’d do with the results.

"We all know what’s been happening here and what’s happened in the past," she said as she watched her class. "I try not to think about it because there's not much I can do."
Who's in charge of genetic testing?

New genetic tests are largely unregulated and presented to an uninformed public.

Is it time for the Food and Drug Administration to step in?
Genetic tests - which can be difficult to perform and even trickier to interpret the results of - have gone largely unregulated by the Food and Drug Administration, by and large leaving commercial biotechnology companies to decide on their own how accurate the tests must be before they're placed on the market.

The FDA has said it has the authority to regulate such tests but has argued in the past that it lacks the resources to provide oversight. Opponents of further oversight worry that beneficial tests would be kept from the public for years.

But a growing apprehension among geneticists and ethicists that a largely uneducated public is being offered tests that may be unproven and unreliable has prompted the FDA to reconsider its role in the marketing of genetic tests.

Last year, the Department of Health and Human Services established an advisory committee to investigate current oversight of the tests.

In March, the committee presented a preliminary report recommending FDA oversight of new tests. In June, following a period of public input, the Secretary's Advisory Committee on Genetic Testing (SACGT) will present its final report.

"What doesn't exist right now is review by the FDA of the validity of the tests," said Sarah Carr, executive secretary of the committee. "The question is, 'Is the current system adequate?'"
Current regulations

Medical experts say there's no way to know the frequency of errors in genetic testing, though it appears to be low. Many patients never find out, and those who do usually settle claims out of court.

With 800 single gene tests currently available, genetic testing can get confusing.

For example, when researchers discovered the genes for breast cancer, scientists worried that promotion of the test would confuse patients - especially those patients who have a family history of breast cancer but not hereditary cancer. For them, the test offered a devastating false hope.

While proficiency testing is mandated for other diagnostic lab tests, it's mostly voluntary for genetic tests.

That issue is one of the gaps the Department of Health and Human Services has tried to examine by establishing SACGT.

The advisory committee is looking at the reliability and quality control standards of genetic testing labs, as well as the training of health care professionals.

Genetic tests may prove particularly difficult to oversee, in part because many physicians aren't adequately trained to interpret the test results or understand when to offer patients a genetic test.

"I know my hands are often tied when trying to convince a physician that a certain test may not be in the best interest of the patient," said Dr. Mary Haag, director of the
A lot of times we’re driven by what patients and physicians are requesting.

“We offer a lot of tests that we wouldn’t recommend in most circumstances,” Haag said.

Shodair can counsel patients about the limitations of a test, but the decision belongs to the patient and their doctor.

Advertising from large pharmaceutical companies may convince some people they want a genetic test that they don’t need. Because the genes of a person are very similar to family members, a test can indicate not only the patient’s risk but an entire family’s risk.

“Testing doesn’t happen automatically [at Shodair], though in some labs it does,” Haag said. “We first make sure the patient has been properly counseled. A positive result can have very damaging effects. Likewise, a negative result can have very damaging effects.”

But adequate counseling is not always available.

Thirty percent of genetics labs don’t have an associated counselor, Haag said. Shodair has three, plus Haag and two physicians.

But for patients who do receive quality care, the benefits of genetic testing are undeniable.
Predictive genetic testing - which measures the risk of developing a disease - can be especially beneficial for patients in high-risk families who test negative and are saved years of worry.

Labs regulated but not the tests

Clinical laboratories should be licensed under the 1988 Clinical Laboratory Improvement Amendments (CLIA), which is regulated by a mix of state, federal and private groups.

"Without compliance with standards already in place, a genetic laboratory doing clinical testing could not get licensed," said Haag of Shodair. "Licensing means credibility."

A survey last spring of 245 molecular genetic testing labs by the Mt. Sinai School Medicine in New York found that 15 percent of the labs scored lower than 70 percent on a quality-control scale.

Standards don't exist for determining which genetic tests are offered to a specific patient, a physician's proficiency at properly interpreting results, or how a sample should be collected and transported.

That becomes important because labs don't offer every available test.
In some cases, a specialized lab may be hundreds or thousands of miles away from where the sample is collected. Shodair has shipped samples as far away as Denmark to test for rare disorders.

Nearly two-thirds of errors in genetic tests occurred in the pretest phase, according to a study published last year in the American Journal of Clinical Pathology. The pretest phase includes determining which test is appropriate, and handling and transportation of the sample.

"I feel there is need to develop oversight," Haag said, "but the testing labs shouldn't be isolated as the only place where this must occur."

**Problems with the proposal**

So how do you test genetic tests? No one seems to know.

Many genetic diseases are very rare, eliminating the opportunity for large-scale studies.

In addition, there aren't many tests to compare a new one to, and even if a test can indicate a mutation, it's difficult to know how often that mutation leads to disease.

At smaller labs, the worry is that new regulations will drive up the cost of testing and staffing, resulting in testing that's concentrated in large commercial labs.
"The people doing the oversight and development of the regulations are not the
ones with the expertise in the field," Haag said. "The regulations they are proposing have
logistic issues that are expensive or impossible."

Smaller labs that specialize or have established testing for rare disorders may be
lost. The result, Haag said, is that the test may no longer be available.

The advisory committee set as one of its goal the continued development of
genetic tests for rare disorders - an endeavor that can be financially draining for a lab if
the test is seldom requested.

The FDA already regulates commercially sold test kits such as home pregnancy or
HIV tests, but unlike simpler blood tests, there are few kits for genetic disorders.

What the FDA doesn't regulate are "home brews" - tests created by an individual
lab for its own use.

While Shodair doesn't develop "home brew" tests, it can send a Montana sample
to a site that has created its own test. Haag says the lab is very cautious about new tests.
 "We look at how good a test is and how good it's been in research," Haag said. "The
question is, 'Can we give the patient something that's worth their money?'"
**Burden of care**

As medicine expands further into prenatal screening and diagnoses, a new legal field has emerged in which parents of children with birth defects hold prenatal providers responsible for failing to detect the problems in utero. Without the tests and the information, parents argue, they can’t make an informed decision whether to abort the child or carry it to term.
Sally and Gaylen Thibeault sued their doctor because their son was born.

Eric Thibeault, now 10 years old, has Down syndrome, a congenital heart problem, and a host of muscular, speech and hearing problems.

Had his mother known of his condition during her pregnancy, she testified in the 1998 lawsuit, she would have aborted him.

But she didn't.

So Sally and Gaylen Thibeault of St. Agatha, Maine, sued their doctor for not identifying their child's problems during the pregnancy – a case they ultimately lost but that raised moral and ethical issues and opened the door for “wrongful birth” lawsuits in the state.

“I hear about cases where people win millions of dollars to care for their kid,” Sally Thibeault said. “We weren’t seeking multi-million dollars – we were seeking to have Eric’s future secured.”

As medicine expands further into prenatal screening and diagnoses, a new legal field has emerged in which parents of children with birth defects hold prenatal providers responsible for failing to detect the problems in utero. Without the tests and the information, parents argue, they can’t make an informed decision whether to abort the child or carry it to term.
"I think patients have a higher expectation of what medicine can achieve," said George Schelling, who represented Dr. Steven Larson, an obstetrician, in the Maine case.

"There was a feeling years ago that that's just the way life is. Now, there clearly is more of a feeling that medicine should be able to achieve just about anything."

When medical providers fail to deliver - or ironically, deliver life - patients are demanding accountability under "wronglife birth":

An Indianapolis girl was born with severe hydrocephaly and died a few months after birth. An ultrasound test showed problems, but a clerical error prevented further testing, and the doctor failed to forward a copy of the ultrasound to the mother's obstetrician. The parents sued, arguing that if they had known of the condition, they would have aborted the child.

A Texas couple was awarded a verdict of $43 million from medical giant Columbia/HCA for refusing the couple's instructions that their severely brain-damaged premature baby not be kept alive through artificial means.

A Massachusetts woman who ran from an abortion clinic in 1994 after a man opened fire said she was so traumatized by the incident that she could not later have an abortion. She sued, arguing that the clinic should pay damages. The woman was at the Preterm Health Services in Brookline in 1994 when John Salvi III began shooting in the lobby, killing a receptionist.
But while "wrongful birth" lawsuits are seen and recognized by many state courts, the cases have rarely, if ever, been seen in Montana.

“It’s certainly a tort which exists,” said Steven Carey, a medical attorney with the Missoula firm Carey, Meismer and McKeon. “But it’s not an area of law that’s going to be developed in Montana – in part because of our small population. Statistically, the lawsuits are a relatively infrequent occurrence.”

Some of the cases are clear-cut, Carey said, and those are likely settled before reaching trial and raising attention in the media.

“To me, it can be a fairly straight forward case,” Carey said. “If through negligence or omission of a test, a health care provider failed to detect a problem with the fetus, you’ve got a valid case for medical negligence.”

In the Thibeault case, jurors had to decide whom to believe.

Dr. Larson, in his second year of practice at the time, twice failed to extract amniotic fluid from Thibeault. He testified Thibeault declined to make the three-hour trip to Bangor, Maine, for another test.

Thibeault said she’d never been informed that there were time constraints on amniocentesis. In Maine, a woman may obtain an abortion up to 24 weeks of gestation. By the time she requested another amniocentesis, that window had passed.
Although there appears to be a legal basis for the cases, many question the ethics of the lawsuits.

Psychologists and health specialists say placing a child in the middle of such cases will likely have a damaging impact on the child’s self-esteem and emotional development.

“If it’s a huge court case with lots of press and attention, then it has the potential to have a major impact on the child,” said Dr. Lynn Montgomery of the Rocky Mountain Perinatal Center. “Depending on the child’s age, they’re going to be able to perceive what’s going on.”

The question is, does a verdict or settlement – which can provide the child with better schooling and health care – make up for the child’s emotional trauma caused by a lawsuit grounded in the notion that the child is a burden to its parents?

Disabled-rights advocates are split by the question, said Michael Guralnick, a child psychologist and executive director at the University of Washington’s Child Health and Human Development Center.

“You have strong advocacy groups who would think that’s the most inappropriate thing to do,” Guralnick said. “There are others that think this would be in the best interest of future disabled people because if you could make people aware of techniques used to maximize a good birth outcome, than this would be in the best interest of children.”
Despite the outcome favoring Larson, the Thibeaults received an unspecified amount of money to help care for their son based on an unusual pretrial agreement.

Jim Marks, director of Disabled Student Services at the University of Montana, suffers from a genetic eye disease that has left him blind. Marks said although money may provide better services for the child, it can’t outweigh the negative effects the trials have on children.

"Is money more important than self-esteem, dignity, and self-determination? Hardly," Marks said. "The funny thing about this argument is that it comes not from the perspective of the person with a disability who respects him or herself. Rather, it comes from another point of view entirely - that of the lawyer who wants the bucks and the parents who leap at the chance to dig into a doctor's deep pockets, no matter the cost of the child's well being."

The cases may affect more than just the individuals involved. Some experts worry that the lawsuits could expand prejudices against people with disabilities, or their parents for having the child.

"The presumption is that the disease is worse than death," Marks said. "[The lawsuits] reinforce the notion that the quality of life is less - that it's a fate worse than death."

Montgomery, a Missoula perinatalologist, said the cases could threaten decades of advance in disabled rights.
"I think we've come so far in taking that stigma away from what a Down syndrome kid is," he said. "To bring it back and make a huge issue of the down sides instead of looking at the potential, it can only put those children back into a bad light."

Doctors, too, may feel the effects of the cases.

"For the most part, I don't think the lawsuits really affect most doctors," Montgomery said. "But it does make some doctors practice more defensively. Maybe they'll order more tests than are really needed."

Marks said he's pleased with advances in prenatal testing, but questioned how some parents are using the knowledge provided by the tests.

Disability, Marks said, is a natural part of life – not the basis of a lawsuit or even a reason to abort a fetus.

"The knowledge [provided by prenatal tests] is not a bad thing," he said. "It's what we do with that knowledge that can be dangerous."
The Philosopher and the Father

*Bioethics:* a discipline dealing with the ethical implications of biological research and applications, especially in medicine.
Fridays with Josh begin late in the afternoon, as the working world goes home to rest.

His father picks him up from the Kent Street Group Home, and the two head to McDonald’s where they order chicken nuggets, two large Cokes and two orders of fries. Always. Doug Cochran-Roberts and his 27-year-old son, born mentally retarded and with severe heart problems, share a passion for the fries.

Four years ago, when Doug and his wife took their son home from the hospital to die, they pulled up to the drive-in window of McDonald’s, even though Josh hadn’t been able to eat for days.

He’d been in the hospital for several weeks, bedridden and nearly comatose after a month-long battle with pneumonia that was killing him in a gradual reduction of life. He had stopped eating. He’d stopped talking. His eyes, his father said, were glazed and distant.

As the family’s van idled at the drive-in window, the smell of the salted fries, that much-maligned cornerstone of fast-food franchises, wafted through the vehicle, and Josh “woke up.”

“He smiled, said his first words and his eyes started to twinkle,” his father said as the two shared a recent Friday meal.

A happy ending? Not really, just a continuation of the life of Josh Roberts.
When Josh was born in the early 1970s, his parents had no idea the extent of his disability. Today, prenatal testing may have told the Robertses of Josh’s condition during the pregnancy, but even with the advances, infants with disabilities often come as a complete surprise to the parents.

Josh recovered from the pneumonia and a list of other ailments, but the seizures didn’t stop, and he’s developed a degenerative brain disease that daily leaves him with a little less function.

“When he recovered,” his father said as Josh chewed on a straw wrapper that he’d mistaken for a french fry, “there was part of me that wished he hadn’t.”

Later, he said:

“For certain families with kids with certain health problems, you begin to wonder if they should continue to live. I don’t think like that very often, but sometimes …”

Meet Peter Singer

Meet Peter Singer: renowned professor at Princeton University’s Center for Human Values, past president of the International Association for Bioethics, and an advocate for euthanization of disabled infants - if the parents choose.

Singer, an Oxford ethicist and Princeton University’s first professor of bioethics, wants to give parents the right to euthanize disabled infants in the first few months of life, a view that has enraged many and fueled a new right-to-life debate.
The stir surrounding Singer stems primarily from his advocacy of allowing parents to euthanize their severely disabled infants – particularly if those infants will experience more pain than pleasure. Singer emphasizes that killing a disabled person who wants to live is never justified, but for disabled infants, he says the choice of life or death should belong with parents and doctors, not state legislators.

Singer includes newborns with disabilities such as spina bifida, categorized by the lack of a completely formed spine, and even hemophilia as those who could be killed if the parents chose. Parents who choose to euthanize their child, he argues, could then attempt to have another, healthier child - one who would experience, and cause, less pain and suffering.

“It’s not as if we’re bringing fewer people into the world,” Singer said in a recent interview. “We’re bringing different people into the world.”

Despite religious or ethical objections, few question the relevancy of Singer’s work. But many question whether his ideas can and will translate into legislation or other practical applications not just in the United States, but around the world.

“I would think quite likely they will,” Singer said. “But I’m really not able to look into the future; I’ve just put forward those views because they seem to me the most humane and compassionate solution to some problems that we do have.

“So it’s not only a matter of does the law change or doesn’t the law change, it’s a matter of the fact that already on an everyday basis hospitals are making decisions about
children with disabilities. I think we need to try and think as clearly as we can about those
decisions.”

The critics’ view

“You’re being a very good boy, Josh Roberts,” Dan Cochran-Roberts said to his
son. Josh said nothing in return. When he does speak, in short, almost violent bursts, his
father often has a difficult time understanding him.

“Josh has had a difficult life,” his father said. “But, you know, just this morning,
my wife said, ‘Most of the time, Josh is happier than we are.”

Josh is tall and thin at 27, looking a dozen years than his age, a teenager waiting
for his body to catch up to his long limbs.

Josh is also the sort of person whom Singer writes about.

But Singer doesn’t live in an academic vacuum.

Anne McDonald, one of Singer’s friends, has athetoid cerebral palsy, a condition
that’s left her wheelchair-bound and reliant on a computerized voice machine to
communicate.

She appears to be precisely whom Singer was referring to in his book Practical
Ethics when he wrote, “Killing a disabled infant is not morally equivalent to killing a
person. Very often it is not wrong at all.”
There’s no paradox here for Singer. He argues that euthanizing such an infant will keep them from suffering, but that for those adults with disabilities, services should be expanded.

“I’m not interested in building a society of perfect people,” Singer said. “We’re never going to get to that point. I’m simply interested in reducing suffering that is pointless.”

Jim Marks, director of Disabled Student Services at the University of Montana, said Singer’s ideas “scare the hell out of me” and remind him of the German medical establishment’s euthanization of people with disabilities during the Holocaust.

“People with disabilities are like a social barometer,” said Marks, who was blinded by a genetic eye disease. “How we are treated indicates the health of a society.”

Critics argue that condoning killing of any kind places humanity on a “slippery slope,” one that could lead to the euthanization of anyone, as all humans experience pain and suffering to an extent.

David Magnus, director of graduate studies at Penn’s Center for Bioethics, said although the ideas leading up to Singer’s conclusions are philosophically sound, the conclusions themselves show a “lack of awareness” of the modern world.

“Personally, I think that there’s a problem with not adjusting your framework to accommodate reasonable conclusions,” Magnus said. “It’s not that unusual to find
philosophers who hold very strange, unintuitive views on the basis of good sound theories."

Glenn McGee, assistant director at Penn's bioethics center, called Singer's work "old news."

"His arguments about disability are incoherent," said McGee, who also writes a monthly bioethics column for MSNBC online. "It is just difficult to imagine a world in which the kind of utilitarianism he proposes would make it into law. People just have more awareness than does Singer that the developing fetus is already a part of our world."

But Singer's views have been gaining attention.

Since his arrival in the United States last fall, he has appeared in Newsweek, The New York Times and The New Yorker among other publications.

His work hasn't been purely academic.

As a student in the 1970s, Singer wrote Animal Liberation, a text that helped create the animal rights movement and converted thousands to vegetarianism.

Part of his work challenged the absence of animals from philosophic considerations. He has argued that newborn humans have no greater right to life than any other species capable of emotion and rationality, including pigs, cows and especially higher primates.

"There isn't any line in intelligence between animals and humans, as such," Singer said. "Some humans with severe intellectual disability are clearly less intelligent
than many non-human animals. That’s really an important part of what I’m saying; that we draw distinctions on the basis of species when we should be looking at individual cases.”

Pleasure and the smell of fries

Josh Roberts loves routine. When he gets to his family’s home he does a head count, making sure everyone’s there for his visit. He sits in his favorite chair and thumbs through magazines, maybe scanning the photos or the words or just the way the lines of black type lie down neatly on top of one another.

No one knows what Josh thinks. But his family feels his effect in every part of their lives, Doug said.

“His mother, sister, cousins – he changes people in a way I never could,” he said.

“If I didn’t have him? … I can’t imagine what I’d be like as a person without him. I just know I’d be less of a human being.”

As for Singer’s arguments, Doug seems torn. Is it okay to love a son so much that you’d let him go, let him die, if it meant less pain for the child? Put yourself at Josh Roberts’ hospital bedside four years ago … or is that even a reasonable request? No one can pretend to know what a parent thinks when they decide to let their son die.
"To make a decision like that, you have to assume so many things that you can’t assume,” Doug said. “What is happiness for my son? What kind of pain lies ahead? We all have pain. Maybe Josh has found within pain a way of seeing the world.”

Later Friday night, father and son get a soda pop at the Two River’s Market in Bonner. The family watches a movie. Josh carries his purple bag to his room and empties it entirely, meticulously arranges its content on the floor, then puts it all back into the pack.

Tomorrow, he’ll rise early, anxious and excited to get back to his friends at the group home. The Friday following, he’ll rise just as early, anxious and excited to get back to his family.

"My son wakes up in the morning and he's excited. He's excited to see his friends, and he's excited to see his family,” Doug said, “and maybe that's a good life."

And in this story, there may be nothing meaningful than this: That pleasure can be as simple as the smell of salted fries.
Interviews

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