

---

## Genetic Services Policy Project

### Cystic Fibrosis: A Vignette

Mark Johnson is a 36-year-old father of two. His daughter, Madison, has cystic fibrosis (CF). Mark and his family live in a small town in Illinois.

Mark first learned about Madison's CF when his wife, Jane, was pregnant. It was their second pregnancy. Their son, Jonathan, was almost 2 years old and healthy. They had briefly discussed carrier screening during their first pregnancy, but decided to forego it because they thought their risk was low. No one in either of their families had CF, though they knew about the condition because a childhood friend of Jane's, Michelle, had had the disease. Michelle had died after a lung transplant several years before, but her younger brother who also had the disease was doing well.

This time when Jane's midwife discussed the recent newborn screening recommendations and offered the CF test, Mark and Jane decided to go ahead with it. The couple was surprised when Jane's test came back positive: she had one copy of the cystic fibrosis gene. They were even more surprised when Mark was also found to be a carrier. At that point, Jane's midwife, in consultation with a local obstetrician, arranged for an amniocentesis (amnio) in Chicago, two hours away. A genetic counselor and a perinatologist told the couple about their 25 percent risk of having an affected child and talked about the options available to them if the test were positive. The amniocentesis was performed and the amniotic fluid sent for DNA analysis.

A week later, the genetic counselor called with the amnio results. Mark and Jane were stunned. They had talked about what they would do if the baby was affected, but were still not expecting the result. Because of their beliefs, as well as Jane's friendship with Michelle, they didn't really consider discontinuing the pregnancy. After the phone call, Mark and Jane drove back to Chicago with a referral to the Cystic Fibrosis Center. The doctors, nurses, and social worker there were particularly helpful in providing information about what to expect. They discussed the challenges of raising a child with CF but were also reassuring about the exciting research being done in this area and the new treatments that were becoming available.

Despite their concerns, once Mark and Jane saw Madison, they knew that things would be okay one way or the other. They were particularly thankful that they knew about the CF before she was born so that they could be prepared, and so she could start enzyme and nutrition therapy immediately after birth. She is now 6 years old and doing so well that, even though Mark doesn't know for sure, he can't help but think the immediate treatment was beneficial. Madison is now in the 50<sup>th</sup> percentile on the growth charts, and is really active. Mark worries that she might get sick at school so he and Jane put her in a private school where they have a little closer connection to the teachers and other parents. They try their best to keep her away from germs, while still trying to lead a pretty normal life.

Currently, Madison is on a special diet, pancreatic enzymes, *recombinant DNase*, and uses a therapy vest two times a day for 30 minutes at a time. Prior to the vest, Mark and Jane were doing chest percussion, which was time- and labor-intensive. For Madison, all the treatments have been a regular part of her life; she doesn't know any differently, though she sometimes wonders why her brother, Jonathan, or other kids don't have to do the same things. The family travels up to the CF Center every three months and has been very happy with the care there. Last time they were there, they talked with the research coordinator about enrolling Madison in a new medication study. At home, Madison is followed by a local pediatrician for routine care. Mark and Jane have been pleased with his coordination of care with the CF Center. Madison has been lucky in avoiding lung infections: she has had no *Pseudomonas* and had only a brief staph infection that resulted in a short hospitalization when she was 3 years old. Other than that incident, she has avoided hospitalizations.

The Johnsons know there are a few other families in nearby communities who have children with CF, but they don't connect with them other than at special events, such as the CF walks. Their main support has been from Michelle's mother, their own family, and online support groups. People (parents and individuals with CF) from all over the country post questions and messages, which has been really helpful. Friends have also been very supportive.

After Mark's positive carrier test, several others in his family got tested too, including his parents. They wanted to know which side of the family the gene was on so that other family members would be aware of their risk. Mark's mother turned out to be a carrier. It is interesting that one of her brothers had trouble with infertility, a possible association with being a CF carrier. His sister hasn't been tested yet. She and her husband are finished having children and have decided to wait until their kids are old enough to choose for themselves whether to be tested. No one in Jane's family wanted testing. Even though their son, Jonathan, didn't have any symptoms of CF, their doctor suggested that Mark and Jane consider testing him since there was a small chance he might have the disease. He was tested and found to be a carrier. Originally, the Johnsons had planned on having three or four children; Jane, especially, wanted a big family. A couple of years after Madison was born, they discussed the possibility of pre-implantation genetic diagnosis. Because of cost and other issues, however, they decided to focus their attention on Madison and Jonathan and possibly consider adoption in the future.

Mark feels that his family has been pretty fortunate. He has a good job and good health insurance. Given the incredibly high cost of CF meds, he can't imagine not having insurance, even though he knows there are assistance programs that can help pay for medicine. He worries at times whether Madison will be able to get insurance coverage when she gets older, a concern he has heard about from the online groups. *Recombinant DNase* costs more than \$1,000 per month, not to mention all the other meds. Even with insurance, the Johnsons pay a significant co-pay on the *recombinant DNase* every month. Madison's therapy vest cost \$16,000. It took over 8 months of filing requests and appealing the insurance company's denials before they finally got it approved for partial coverage.

Mark is amazed that scientists know so much about the gene for cystic fibrosis, but don't have a cure yet. He and Jane decided to contribute to a national cystic fibrosis research fund to help find

**Genetic Services Policy Project**  
<http://depts.washington.edu/genpol>

a cure. They are hopeful for the future of gene therapy and other treatments, but in the meantime, they are glad for the technology that allowed them to identify Madison's CF so early. After talking with Michelle's mother about all the hurdles they went through before Michelle was diagnosed with CF 25 years ago at age 4, Mark has become an advocate for screening newborns for CF. Illinois was one of the states that did not screen newborns for cystic fibrosis, so when he was asked to serve on the state newborn screening advisory committee, Mark readily agreed. He is excited that the state has now decided to add CF to its screening panel. He knows there is some controversy about the value of screening all babies for CF, but he thinks if screening can find just one child with CF, it will be worth it.

***Genetic Services Policy Project***  
<http://depts.washington.edu/genpol>

This work is supported in part by Projects # U35MC02601 and # U35MC02602 from the Maternal and Child Health Bureau (Title V, Social Security Act), #11223, Health Resources and Services Administration, Department of Health and Human Services.

## Case Issues for Discussion

1. The couple in this case chose to undergo carrier screening for the cystic fibrosis gene after the midwife discussed new screening recommendations from a professional organization. In addition to offering prenatal carrier screening to all couples, the recommendations also suggest that screening should be offered to couples planning a pregnancy. Studies indicate that while prenatal screening is being widely implemented, preconception screening is still limited.
  - a. What are the potential benefits and harms of preconception carrier screening?
  - b. What barriers exist to implementing preconception screening recommendations in practice?
  - c. What opportunities exist to overcome these barriers?
  
2. After learning that their unborn baby had CF, Mark and Jane were faced with a choice regarding continuing or terminating the pregnancy.
  - a. What factors played into their decision?
  - b. What supports would help Mark and Jane make their decision?
  
3. Madison is doing well overall.
  - c. How might this scenario differ if she was experiencing more significant health problems?
  - d. How might the scenario differ if her family situation was different (e.g., single parent, language or cultural barriers, limited education, lack of a job or health insurance, lack of geographic access to CF services)?
  
4. As demonstrated in this case, multiple providers are involved in Madison's care. Coordination of care is a significant issue, particularly in areas that do not have a specialty care center.
  - a. What systems and resources are needed to assure a close connection between community primary care providers and specialty care providers based in the CF center?
  - b. What are the potential problems if care is not well coordinated?
  - c. What educational needs do each of the various providers have?
  - d. What is the role of the medical home?
  - e. What is the role of the patient and family?
  
5. In the United States, the development of accredited cystic fibrosis specialty care centers has been touted as a model for other chronic diseases. Currently (as of 2007), there are 115 accredited centers, 95 adult programs, and 50 affiliated centers nationwide.
  - a. What are the advantages of this model of care?
  - b. What are the disadvantages?
  - c. What role does accreditation play in assuring quality of care?
  
6. Treatment for CF is costly and often includes "experimental" therapies or procedures that may or may not be covered by an individual health plan.
  - a. What is the impact of insurance coverage decisions on individuals and families with CF?
  - b. What is the role of public assistance programs?
  - c. What is the role of private assistance programs (e.g., pharmaceutical assistance programs, foundation programs)?

- d. Why do adults with CF have particular problems with insurance coverage?
  - e. What resources or services are available for adults?
  - f. Some states (e.g., Florida) have considered legislation to mandate insurance coverage of CF treatments and services. What are the pros and cons of such legislation? Why might insurance companies oppose such mandates?
7. Research is a significant component of CF care. The family in this case is considering enrolling Madison in a medication research study.
- a. What, if any, ethical concerns are associated with research in children? What protections would address these concerns?
  - b. Should risky experimental treatments (e.g., gene therapy) be withheld from children, even if there are potentially lifesaving benefits?
8. As depicted in this scenario, different family members make different choices about genetic testing.
- a. What are the indications for testing family members?
  - b. What, if any, ethical concerns are associated with testing children for carrier status?
9. The couple has considered having additional children.
- a. What is the role of pre-implantation genetic diagnosis?
  - b. What ethical dilemmas are associated with this technology for this condition and others?
10. Advocacy and political action have played significant roles in raising the profile of this disease, as well as attracting financial support for the cause.
- a. What are the pros and cons of advocacy group involvement in policy development?
  - b. What mechanisms would contribute to equitable allocation of resources for various diseases and causes?
11. Mark mentions the controversy surrounding screening newborns for cystic fibrosis.
- a. Why is universal newborn screening for CF controversial?
  - b. What are the arguments for newborn screening for CF?
  - c. What are the arguments against it?

What service delivery issues does this scenario raise?

- Geographic access to services
- Specialty care center model

What provider issues are identified?

- Multiple providers (including prenatal, pediatric, specialty)
- Coordination of care
- Implementation of professional recommendations and clinical guidelines
- Genetic testing: who and when to test

What consumer issues are identified?

- Difficult choices/ethical dilemmas related to choices
- Financial concerns: cost-sharing and out-of-pocket expenses
- Consumer advocacy/involvement in policy making bodies
- Ethical concerns about research in children
- Family issues

What payer or coverage issues are identified?

- Consumer cost sharing
- Impact of coverage decisions on consumers
- Coverage of new technologies and “experimental” treatments
- Role of public and private financial assistance programs

What industry issues are identified?

- Development and role of new treatments (e.g., gene therapy)

What policy issues are suggested?

- Newborn screening