Facioscapulohumeral muscular dystrophy (FSHD) is the second most common adult muscular dystrophy with an estimated prevalence of 1:20,000. The disease is dominantly inherited and linked to a deletion of variable size in a 3.3-kb repetitive DNA sequence on chromosome 4q35. A causative gene has not been identified, and the pathophysiology of the disease remains unclear. It occurs equally in males and females, but women tend to be less severely affected than men. Disease progression is usually slow, and life expectancy is normal, although about 20% of patients become wheelchair bound.1,2 Most cases do not report complications such as worsening muscle weakness during pregnancy, with the possible exception of some women who are wheelchair bound.3

Methods. In this cross-sectional study, subjects were recruited through a mail-out announcement letter to all women with FSHD enrolled in the National Registry of Myotonic Dystrophy and FSHD patients and family members (n = 186). Additional subjects were recruited from respondents to advertisements posted through the FSH Society and Muscular Dystrophy Association. A questionnaire was mailed to those women who elected to participate. Approximately 30% of women did not participate, and 73% completed the questionnaire. Additional questionnaires were returned by telephone interviews carried out when necessary. The information from the questionnaires was combined with the review of medical records. Pregnancy outcomes were compared to published national data from the Centers for Disease Control and Prevention4,5 with χ² analysis.

Results. Complete questionnaires were returned by 48 women. Education level, employment status, age, and geographic location did not differ between responders and nonresponders in the National Registry. Ten women never had a pregnancy, and six of them decided so because of having FSHD. Thirty-eight women reported a total of 105 gestations (range per woman one to six, mean 2.7) and 78 live births. The diagnosis of FSHD was genetically confirmed in 26, clinically definite in eight, and clinically probable in four. Mean maternal age at the time of gestation was 28.3 (range 17 to 40, SD = 5.15). The diagnosis of FSHD at the time of gestation was known in 46% of cases. The majority of women rated their functional disability from FSHD at the time of pregnancy as mild (63%) or moderate (37%). Only four women used an assistive device during their pregnancies: one woman used a wheelchair (two pregnancies), two women used an ankle brace (three pregnancies), and one used a scooter and a walker (one pregnancy).

Pregnancy and birth complications are shown in the table. Cesarean delivery was more common in these patients with FSHD than in national birth data, even when taking into account the variation in cesarean delivery rate over the time frame of the births reported (figure). If only primary cesarean delivery is examined, there is no difference between the patients who responded to our questionnaire and national data from the same years.

The rate of forceps deliveries is markedly higher than Obstetric risk in facioscapulohumeral muscular dystrophy (FSHD) is not known. We surveyed 38 women with FSHD reporting 105 gestations and 78 live births. Review of medical records showed that pregnancy outcomes were generally favorable. The rates for low birth weight and total operative deliveries were statistically higher than the national rates in the general population. Worsening of FSHD was reported in 24% of gestations and did not usually resolve after delivery.
reported in national data, but the rate of vacuum-assisted vaginal deliveries is the same. The combined rate for all operative vaginal deliveries is significantly higher in this sample of patients with FSHD than in the general population. The rate of forceps deliveries was also higher in the group who did not know they had FSHD.

The incidence of prematurity did not differ from the general population. However, a significantly higher incidence of low birth weight infants was noted. This was not associated with a higher incidence of preeclampsia, other pregnancy complications, or neonatal death.

Although women reported higher rates of fetal distress, infection, and anesthetic complications on their questionnaires than occur in the general population, review of the obstetric records did not confirm these findings. In the records available, the incidence of both fetal distress and obstetric infection was not different than the national rates. Adequate records were not available to assess anesthetic complications.

Of the 105 gestations, 24% resulted in worsening of FSHD symptoms that for the most part did not resolve after childbirth. The most common complaints were, in order of frequency, worsening of generalized weakness, frequent falling, difficulty carrying the infant due to worsening of shoulder weakness, worsening or new-onset pain, and difficulty carrying the infant due to worsening of leg weakness. Of the 38 women who became pregnant, 90% reported that they would choose pregnancy again, and many added comments to this effect on the questionnaire.

Discussion. Overall, pregnancy outcomes in patients with FSHD were good. Our study shows an increased incidence of operative vaginal delivery in women with FSHD despite the fact that the majority were mildly or moderately affected. Moreover, this finding cannot be attributed to bias in favor of surgical delivery because of the diagnosis as the diagnosis of FSHD was not known at the time of the majority of the deliveries. This suggests compromise of the second stage of labor when skeletal muscle effort is required and is likely due to the typical abdominal and truncal muscle weakness very commonly associated with FSHD even in the early stages.

The significantly higher rate of low birth weight is unexpected and difficult to interpret based on what is known about the pathophysiology of FSHD. Further evaluation of this pregnancy outcome is warranted. In addition, the possible association between FSHD and obstetric anesthetic complications should be investigated.

Exacerbation of muscle weakness and pain was reported in 24% of the women, similar to what was found in a previous report and consistent with what many women in our neuromuscular clinic have reported anecdotally. The women in our group denied resolution of their symptoms after childbirth, suggesting a possible irreversible effect of pregnancy on the progression of the disease.

Limitations of this study include information from a self-selected group and possible recall bias, but the data are nevertheless important given the paucity of information currently available. Our sample appears to be representative of the spectrum of disease severity seen in FSHD.

Discussion. Overall, pregnancy outcomes in patients with FSHD were good. Our study shows an increased incidence of operative vaginal delivery in women with FSHD despite the fact that the majority were mildly or moderately affected. Moreover, this finding cannot be attributed to bias in favor of surgical delivery because of the diagnosis as the diagnosis of FSHD was not known at the time of the majority of the deliveries. This suggests compromise of the second stage of labor when skeletal muscle effort is required and is likely due to the typical abdominal and truncal muscle weakness very commonly associated with FSHD even in the early stages.

The significantly higher rate of low birth weight is unexpected and difficult to interpret based on what is known about the pathophysiology of FSHD. Further evaluation of this pregnancy outcome is warranted. In addition, the possible association between FSHD and obstetric anesthetic complications should be investigated.

Exacerbation of muscle weakness and pain was reported in 24% of the women, similar to what was found in a previous report and consistent with what many women in our neuromuscular clinic have reported anecdotally. The women in our group denied resolution of their symptoms after childbirth, suggesting a possible irreversible effect of pregnancy on the progression of the disease.

Limitations of this study include information from a self-selected group and possible recall bias, but the data are nevertheless important given the paucity of information currently available. Our sample appears to be representative of the spectrum of disease severity seen in FSHD.

Discussion. Overall, pregnancy outcomes in patients with FSHD were good. Our study shows an increased incidence of operative vaginal delivery in women with FSHD despite the fact that the majority were mildly or moderately affected. Moreover, this finding cannot be attributed to bias in favor of surgical delivery because of the diagnosis as the diagnosis of FSHD was not known at the time of the majority of the deliveries. This suggests compromise of the second stage of labor when skeletal muscle effort is required and is likely due to the typical abdominal and truncal muscle weakness very commonly associated with FSHD even in the early stages.

The significantly higher rate of low birth weight is unexpected and difficult to interpret based on what is known about the pathophysiology of FSHD. Further evaluation of this pregnancy outcome is warranted. In addition, the possible association between FSHD and obstetric anesthetic complications should be investigated.

Exacerbation of muscle weakness and pain was reported in 24% of the women, similar to what was found in a previous report and consistent with what many women in our neuromuscular clinic have reported anecdotally. The women in our group denied resolution of their symptoms after childbirth, suggesting a possible irreversible effect of pregnancy on the progression of the disease.

Limitations of this study include information from a self-selected group and possible recall bias, but the data are nevertheless important given the paucity of information currently available. Our sample appears to be representative of the spectrum of disease severity seen in FSHD.
Locked-in syndrome resulting from bilateral cerebral peduncle infarctions

Tarek Zakaria, MD; and Matthew L. Flaherty, MD, Cincinnati, OH

A 71-year-old man had acute onset of ataxia, dysarthria, and visual blurring, which progressed in a stuttering fashion to a locked-in syndrome. Only pupillary reflexes and extraocular movements (both vertical and lateral) were ultimately preserved.

Figure. (A) Diffusion-weighted MRI sequence shows acute infarction of the cerebral peduncles. (B) Intracranial MRA. The basilar artery and posterior cerebral arteries (PCAs) appear occluded on the reconstructed image, but source images suggest sluggish PCA flow from posterior communicating arteries.

Disclosure: The authors report no conflicts of interest.

Address correspondence and reprint requests to Dr. Matthew L. Flaherty, 231 Albert Sabin Way, MSB Room 5161B, University of Cincinnati Medical Center, Cincinnati, OH 45267-0525; e-mail: matthew.flaherty@uc.edu

Copyright © 2006 by AAN Enterprises, Inc.

Locked-in syndrome resulting from bilateral cerebral peduncle infarctions
Tarek Zakaria and Matthew L. Flaherty
*Neurology* 2006;67;1889
DOI 10.1212/01.wnl.0000229160.49552.b0

This information is current as of November 27, 2006

<table>
<thead>
<tr>
<th>Updated Information &amp; Services</th>
<th>including high resolution figures, can be found at: <a href="http://www.neurology.org/content/67/10/1889.full.html">http://www.neurology.org/content/67/10/1889.full.html</a></th>
</tr>
</thead>
<tbody>
<tr>
<td>Supplementary Material</td>
<td>Supplementary material can be found at: <a href="http://www.neurology.org/content/suppl/2007/05/31/67.10.1889.DC1">http://www.neurology.org/content/suppl/2007/05/31/67.10.1889.DC1</a></td>
</tr>
<tr>
<td>References</td>
<td>This article cites 2 articles, 0 of which you can access for free at: <a href="http://www.neurology.org/content/67/10/1889.full.html##ref-list-1">http://www.neurology.org/content/67/10/1889.full.html##ref-list-1</a></td>
</tr>
<tr>
<td>Subspecialty Collections</td>
<td>This article, along with others on similar topics, appears in the following collection(s):</td>
</tr>
<tr>
<td></td>
<td><strong>All Cerebrovascular disease/Stroke</strong> <a href="http://www.neurology.org/cgi/collection/all_cerebrovascular_disease_stroke">http://www.neurology.org/cgi/collection/all_cerebrovascular_disease_stroke</a></td>
</tr>
<tr>
<td></td>
<td><strong>Coma</strong> <a href="http://www.neurology.org/cgi/collection/coma">http://www.neurology.org/cgi/collection/coma</a></td>
</tr>
<tr>
<td></td>
<td><strong>Infarction</strong> <a href="http://www.neurology.org/cgi/collection/infarction">http://www.neurology.org/cgi/collection/infarction</a></td>
</tr>
<tr>
<td></td>
<td><strong>MRI</strong> <a href="http://www.neurology.org/cgi/collection/mri">http://www.neurology.org/cgi/collection/mri</a></td>
</tr>
<tr>
<td>Permissions &amp; Licensing</td>
<td>Information about reproducing this article in parts (figures, tables) or in its entirety can be found online at: <a href="http://www.neurology.org/misc/about.xhtml#permissions">http://www.neurology.org/misc/about.xhtml#permissions</a></td>
</tr>
<tr>
<td>Reprints</td>
<td>Information about ordering reprints can be found online: <a href="http://www.neurology.org/misc/addir.xhtml#reprintsus">http://www.neurology.org/misc/addir.xhtml#reprintsus</a></td>
</tr>
</tbody>
</table>

*Neurology* © is the official journal of the American Academy of Neurology. Published continuously since 1951, it is now a weekly with 48 issues per year. Copyright. All rights reserved. Print ISSN: 0028-3878. Online ISSN: 1526-632X.