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

There is limited information currently available to guide obstetricians and neurologists in counseling women with FSHD who are planning to become pregnant and give birth.3 The possible effect of pregnancy on the progression of muscle weakness is unknown.

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 and signed consent and release of medical information forms (n = 73). The questionnaire was designed to capture information regarding the decision to become pregnant, genetic counseling, severity of FSHD, pregnancy outcome, type of delivery, postpartum complications, worsening of muscle weakness during pregnancy, and difficulty caring for the newborn. Self-reported assessment of FSHD severity as mild, moderate, or severe was matched to their response to questions regarding functional limitations at the time of pregnancy. FSHD diagnosis was confirmed in each case through medical records review and defined using the three classifications. Genetically confirmed was defined as having weakness consistent with FSHD and a positive FSHD genetic test (allele size \( \leq 38 \) kb) in the subject or an affected relative. Clinically definite was defined as muscle weakness in the face in addition to either the scapular stabilizers or foot dorsiflexors, no eyelid or extracranial muscle weakness, a dominant family history, EMG, or muscle biopsy not suggestive of alternative diagnosis, and genetic testing not done. Clinically probable was defined as muscle weakness in the face plus either scapular stabilizers or foot dorsiflexors, no eyelid or extracranial muscle weakness, EKG, and muscle biopsy not done or reports unavailable, and genetic testing not done.

All completed questionnaires were reviewed for accuracy, and telephone interviews were carried out when necessary. The information from the questionnaires was combined with the review of medical records from obstetricians and neurologists caring for the patients at the time of pregnancy. The study was approved by the University of Rochester Institutional Review Board. Pregnancy outcomes were compared to published national data from the Centers for Disease Control and Prevention4,5 with \( \chi^2 \) 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 \( \pm 5.15 \)). 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...
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.

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References

NeuroImages

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.

Magnetic resonance imaging (MRI) showed bilateral infarctions of the cerebral peduncles with sparing of the thalami and occipital lobes (figure). The basilar terminus and posterior cerebral arteries (PCAs) appeared occluded on the reconstructed MRA, but source images suggested sluggish PCA flow via posterior communicating arteries, potentially explaining the limited distribution of infarction. Locked-in syndrome usually results from ischemic, traumatic, toxic, or demyelinating lesions of the ventral pons. It is rarely caused by infarction of the cerebral peduncles, which are supplied by multiple perforating arteries originating from the PCAs.1,2

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.

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Locked-in syndrome resulting from bilateral cerebral peduncle infarctions

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