OBSTETRICS

Pregnancy with Friedreich ataxia: a retrospective review of medical risks and psychosocial implications

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OBJECTIVE: Friedreich ataxia (FRDA) is an autosomal recessive, neurodegenerative disease. Recent medical advances have improved the average life expectancy, and as such, many female patients are contemplating pregnancy. However, little research exists exploring the medical or psychosocial complications that arise from pregnancy with this disease.

STUDY DESIGN: In this study, we retrospectively examined 31 women with FRDA who had 65 pregnancies, resulting in 56 live offspring.

RESULTS: FRDA did not appear to increase the risk of spontaneous abortion, preeclampsia, or preterm birth. Despite the sensory and proprioceptive loss that occurs in FRDA, nearly four fifths of births were vaginal. Of babies, 94.4% were discharged home with their mothers. Equal numbers of women reported that pregnancy made their disease symptoms worse, better, or unchanged.

CONCLUSION: This study demonstrates that women with FRDA can have uncomplicated pregnancies that do not uniformly complicate disease symptomatology.

Key words: autosomal recessive genetic disease, family planning, Friedreich ataxia, neurodegenerative disease, pregnancy with disability

Cite this article as: Friedman LS, Paulsen EK, Schadt KA, et al. Pregnancy with Friedreich ataxia: a retrospective review of medical risks and psychosocial implications. Am J Obstet Gynecol 2010;203:224.e1-5.

Friedreich ataxia (FRDA) is a neurodegenerative, autosomal recessive genetic disorder characterized by dysarthria, ataxia, loss of reflexes, scoliosis, cardiomyopathy, and balance difficulties.[1-4] Typical onset occurs in late childhood or early adolescence, but is variable.[3] Of people with FRDA, 97% have an expanded GAA triplet repeat in both alleles of the *FXN* gene, and the length of the shorter GAA repeat correlates with age of onset ($r = 0.6$-7).[5-7] The remaining 3% of patients carry an expanded GAA repeat on one allele, and a point mutation on the other allele.[8-10]

With improved cardiac care, spinal fusion surgery, and other medical interventions, the mean life expectancy of FRDA has likely improved significantly beyond the previously reported age of 37 years.[11] As a result, a significant number of women with FRDA are contemplating pregnancy. Many are seeking specific guidance on family planning, an issue that has not been adequately addressed.

Few studies have examined disease-associated medical risks and/or psychosocial implications of pregnancy in women with FRDA. One report detailed the outcomes of 17 pregnancies in women with FRDA, and identified pregnancy-induced hypertension as a complication in 11.8% of that cohort.[12] Another case report of a woman with FRDA reported the development of profound weakness and respiratory distress, secondary to the administration of magnesium sulfate to control preterm labor and preeclampsia.[13] In the present study, we retrospectively examined 31 women with FRDA who experienced 65 pregnancies resulting in 56 live offspring.

Materials and Methods

This study had the approval of the institutional review boards of our hospital and university. All subjects provided written informed consent before taking part in the study. The study was open to any woman with FRDA age >18 years who had at least 1 pregnancy, including those that ended in miscarriage. Subjects were recruited through our FRDA program and by word of mouth. At the time of study recruitment, there were roughly 140 patients, approximately equally divided by gender, followed up through the program.

We retrospectively interviewed 33 women; 2 women were excluded from data analysis because their diagnosis of FRDA could not be confirmed through medical documentation. Each subject filled out a questionnaire detailing her experience with pregnancy. The questionnaire focused on 2 overall aspects: (1) medical concerns and complications associated with pregnancy; and (2) psychosocial factors influencing the decision to have a baby. Specific questions detailed past and present medical his-

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Received Dec. 31, 2009; revised Feb. 25, 2010; accepted March 22, 2010.

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This study was funded by a grant from the National Ataxia Foundation.

0002-9378/$36.00
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doi: 10.1016/j.ajog.2010.03.046

TABLE 1
Cohort demographics at time of study participation

Characteristic	Women
Married	84%
Children reside with mother full time	77%
Reside on East Coast	52%
Reside with partner	88%
Perform all or most childcare	55%
Unemployed	61%
Nonambulatory	61%

Friedman. Pregnancy with Friedreich ataxia. Am J Obstet Gynecol 2010.

tory, age of FRDA diagnosis, history of pregnancies and deliveries, and feelings about pregnancy relating to FRDA. The questionnaire included both open- and closed-ended questions, as well as ranked items employing the Likert scale.

Two versions of the questionnaire were administered: one for women who were diagnosed with FRDA prior to their first pregnancy, and a second for women diagnosed after their first pregnancy. The questionnaires were nearly identical, except the former asked questions about how FRDA influenced the decision to become pregnant, and the latter asked questions about changes the women may have made knowing the FRDA diagnosis before pregnancy. For women unable to complete the questionnaire (for reasons such as inability to write), the study coordinator administered the questionnaire verbally. Obstetric medical records were obtained and reviewed for 19 of the women whose data are included in this study. For women with children age >18 years, it was not possible to obtain medical records from these pregnancies or births.

TABLE 2
Commonly reported health problems at time of study participation

Health problem	Women
Scoliosis	48.4%
Urinary incontinence	25.8%
Left ventricular hypertrophy	22.6%
Depression or anxiety	22.6%
Hypercholesterolemia	9.7%
Hypothyroidism	9.7%
Diabetes type 2	9.7%
Hypertension	6.5%
Diabetes type 1	3.2%
Ulcerative colitis	3.2%

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RESULTS

Cohort demographics

In all, 31 women were included in data analysis, all of whom resided in the United States and carried a confirmed clinical diagnosis of FRDA. Thirteen women (42%) were seen in the FRDA program. In the overall cohort, the mean length of the shorter GAA triplet repeat in the *FXN* gene was 421 repeats, with a minimum of 44 repeats and a maximum of 760 repeats. Two women carried point mutations (G130V and I154F). The average age of FRDA diagnosis was 24.4 years, and the average age at study participation was 38.2 years (Table 1). A total of 21 women (68%) were diagnosed with FRDA prior to their first pregnancy. The women had an average of 1.8 children each. The women also had a variety of health problems associated with FRDA[1,4,14] such as scoliosis, urinary incontinence, and left ventricular hypertrophy (Table 2).

Pregnancy data

In all, 31 women had a total of 65 pregnancies, including 1 twin gestation. Of those pregnancies, 55 resulted in 56 live-born children. Nine pregnancies (13.8%) ended in spontaneous abortion (SAB), a smaller percentage than the estimated national incidence.[15] Of that group, 4 women each experienced a SAB, 1 woman had 2 SABs, and 1 woman had 3 SABs. These occurred at an average of 8.17 weeks of gestation. There was no information in any of the medical records to suggest that the SABs were related to FRDA, and all the women had other term pregnancies. Additionally, a 36-year-old woman terminated a pregnancy at 16 weeks, following prenatal diagnosis of Trisomy 18, presumptively attributed to advanced maternal age.[16] This mother had no further pregnancies.

The average age of the cohort during first pregnancy was 25.7 years. In all, 22 women (70.9%) planned their first pregnancy. Of that group, the average time to get pregnant was 97.2 days. Eleven women (35.5%) requested their partner undergo FRDA carrier testing prior to conception. All of the women received regular obstetric care during pregnancy, with 4 (6.2%) pregnancies deemed high risk, 1 of which was considered high risk due to FRDA. In this case, the mother was nonambulatory and her physician believed she was at risk for the development of deep vein thrombosis. Thirteen women (41.9%) were followed up by a cardiologist during pregnancy. One woman was a type 1 diabetic prior to pregnancy and 1 woman developed gestational diabetes, which occurs in 4-8% of pregnancies.[17,18] Four women (12.9%) had abnormal glucose tolerance test results during pregnancy, but did not require pharmacologic intervention. For the 11 women on whom data were available, the average weight gain during pregnancy was 36.6 lb.

The women were asked to retrospectively rate if they believed pregnancy made the FRDA worse, better, or unchanged. Eight women (28.6%) believed pregnancy improved their symptoms, typically citing a feeling of improved balance and coordination. Ten women (35.7%) believed pregnancy made their symptomatology worse, most commonly experiencing increased fatigue, followed by urinary urgency, and further speech, balance, and coordination difficulties. The remaining women believed pregnancy did not alter their symptoms of FRDA.

Thirty women carried pregnancies to term, and of those, 20 women (66.7%) reported experiencing at least 1 complication during pregnancy (Table 3). The most common complication was preterm birth, identified in 12.96% of FRDA pregnancies. This rate is similar to the general population incidence.[19] Of those who experienced complications, 18 women (90%) believed them to be unrelated to FRDA.

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