FSHD and Pregnancy

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FSHD affects women in their childbearing years and muscle weakness can typically target the abdominal wall and back muscles. The severity of muscle weakness and the age of onset are variable. Many women with FSHD (more than 50%) don't know about their diagnosis at the time of their pregnancy.

Limited information is currently available about pregnancy and birth outcomes in FSHD and the potential effect of pregnancy and menopause on the progression of FSHD is largely unknown. The currently available data suggest that overall pregnancy outcomes in FSHD are very good.

Will the disease affect fertility?

- This has not been systematically studied in FSHD but there is no reported evidence to suggest that fertility is affected

Are there specific risks to the fetus?

- The incidence of prematurity (babies born before 37 weeks), fetal distress, or neonatal death in babies born to mothers with FSHD does not differ from the general population
- There is a significantly higher incidence of low birth weight (birth weight less than 2500 grams) in babies born to mothers with FSHD

- Each baby will have a 50/50 chance to inherit FSHD since this disease is almost always autosomal dominant in inheritance

Will the disease affect the ability to carry a pregnancy?

- No, miscarriage and preterm labor are not increased in FSHD

Are there more obstetric complications during pregnancy and delivery?

- C-Sections and forceps deliveries are increased in FSHD. This could be due to the weakness in the abdominal wall muscles.
- There is no increased risk for preeclampsia, polyhydramnios, premature rupture of the membranes, gestational diabetes or birth defects.

Is there a greater risk if anesthesia is used during delivery?

- There is no data to suggest increased risk with general anesthesia

Will the course of FSHD be affected by pregnancy?

- In 1 out of 4 women pregnancy results in worsening of FSHD symptoms that for the most part does not resolve after childbirth.
- Most common problems are: worsening of generalized weakness, frequent falling, difficulty carrying the baby because of worsening shoulder weakness or leg weakness, worsening or new onset pain.
- Most women (90%) would choose pregnancy again

Will the course of FSHD be affected by menopause?

- This is unknown at this time and further research is needed

Recommendations for Women with FSHD and Their Providers at the time of Pregnancy and Delivery

- **Before Pregnancy**: Woman with FSHD or with family history of FSHD who are planning a family should consider genetic testing if not already done to confirm or rule out the diagnosis and to look for certain genetic test results such as the size of the D4Z4 deletion that may suggest more significant disability at an earlier age. Genetic counseling should be considered to discuss reproductive options: having children without testing, pre-implantation genetic diagnosis, prenatal screening, gamete donation, adoption.

- **During Pregnancy**: Women with FSHD who are pregnant should be assessed for maternal risk factors, obstetric and FSHD –specific: 1) Respiratory function (Forced Vital Capacity sitting and supine) should be assessed at baseline and throughout the pregnancy and especially in the third trimester when weight gain can affect the diaphragmatic function especially when supine; 2) while routine cardiac screening is not necessary in FSHD patients, cardiac evaluation should be obtained if patients develop signs and symptoms suspicious for heart disease (i.e. shortness of breath, palpitations, chest pain); 3) assess and manage pain; 4) Physical therapy evaluation to assess the degree of arm and shoulder weakness, ambulation status, need for

adaptive equipment/bracing, and change in functional status throughout pregnancy; 5) consider a consult with a dietician to assess BMI at baseline and to discuss target weight gain for pregnancy. Excessive weight gain and change in the center of gravity during pregnancy may increase falls and precipitate loss of the ability to walk independently.

The patient's neuromuscular neurologist should help educate and coordinate care with the obstetrician and the other specialists involved in the pregnancy and delivery. Remember, FSHD is a relatively rare disease and not all providers are familiar with the specifics aspects of FSHD. Knowledgeable patients are healthier patients!

- **Labor and Delivery**: The mode and place of delivery (tertiary center or local hospital) and the choice and dosing of pain medications should be discussed in the third trimester in a multidisciplinary team meeting where all medical information can be reviewed and discussed by the patient's neurologist/neuromuscular specialist, obstetrician, anesthesiologist, and pulmonologist if indicated. If a vaginal delivery is planned, the team should be prepared for possible assisted or operative delivery.
  Having FSHD is not in itself an indication to have a C section and C section should be planned only for obstetrics indications (large baby, baby position, etc.), or if indicated based on severe degree of FSHD weakness or respiratory insufficiency or other medical comorbidities.
  The first stage of labor should not be affected by FSHD. In the second stage of labor, weak skeletal abdominal wall muscles frequently affected in FSHD

may cause slowing in labor progression and need for assisted delivery with vacuum or forceps. Epidural anesthesia is generally preferred in neuromuscular diseases.

- **After delivery**: Advocate for longer inpatient stay to allow for assessment and monitoring of deterioration of motor function and pain. Consider physical and occupational therapy and lactation consults: caring for a new baby is an activity of daily leaving and the need for assistive devices and adaptive equipment should be considered and reassess as motor function might have change with pregnancy and delivery. Monitor for post-partum depression: women with functional limitations are more likely to develop post-partum depression than women without disabilities

References

Ciafaloni *et al.* "Pregnancy and birth outcomes in women with facioscapulohumeral muscular dystrophy". Neurology 2006; 67: 1887-1889.

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Evidence-based guideline summary: Evaluation, diagnosis, and management of facioscapulohumeral muscular dystrophy: Report of the Guideline Development, Dissemination, and Implementation Subcommittee of the American Academy of Neurology and the Practice Issues Review Panel of the American Association of Neuromuscular & Electrodiagnostic Medicine 2015