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.

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.

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.

Rudnick-Schoneborn *et al*. "Obstetric aspects in women with facioscapulohumeral muscular dystrophy, limb-girdle muscular dystrophy, and congenital myopathies". Arch Neurol. 1997; 54(7):888-94.

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