



Reproductive System

Symptoms

Testicular Atrophy

Primary hypogonadism in males (testicular atrophy) is usually not recognized until adulthood. Symptoms can include:

- Small testes, associated with decreased or absent sperm production. Infertility issues are more common in patients with DM1.
- Weak secondary sex characteristics, including decreased energy, libido, sexual hair, muscle mass, and bone mineral density
- Low serum testosterone (low or low-normal urinary 17-ketosteroid (17-KS) excretion, prohormone precursors of testosterone and estrone/estradiol)
- Elevated serum FSH and LH concentration
- Elevated FSH levels can result in high estradiol:testosterone ratios, leading to gynecomastia

Female Infertility

- Reduced fertility is seen in females with myotonic dystrophy, however there is little evidence of gonadal dysfunction or hypogonadism. Infertility symptoms include:
 - Increased spontaneous abortion and stillbirth rate
 - Early menopause in rare cases

Pregnancy Complications

Maternal complications during pregnancy may include:

- Prolonged labor and delivery related to uterine dysfunction, maternal weakness, and lack of voluntary assistance
- Uterine overdistention, related to polyhydramnios, which can cause preterm labor, inadequate uterine contractions (atonic uterus), or premature spontaneous rupture of membranes
- Myotonic spasms following the administration of depolarizing agents; respiratory depression following the administration of barbiturates
- Post-partum hemorrhage due to inadequate uterine contractions (atonic uterus) or retained placenta

Neonatal Complications

Fetal and neonatal complications in newborns with congenital myotonic dystrophy type 1 (DM1) may include:

- Polyhydramnios, which is associated with increased risks of adverse pregnancy outcome
- Umbilical cord prolapse or placental abruption



- Fetal malposition due to reduced fetal mobility
- Pre-term labor
- Hydrops fetalis
- Fetal akinesia

Diagnosis

Reproductive issues

Diagnosis of fertility issues of individuals (males and females) with myotonic dystrophy may include:

- Blood tests to measure circulating hormone levels (including testosterone, estradiol, FSH, LH, and thyroid hormones)
- Semen analysis (where possible)

Pregnancy Complications

Polyhydramnios is typically diagnosed by ultrasound examination. An increase in amniotic fluid volume may be qualitative or quantitative. Serial examinations can identify potential issues, even if sensitivity and positive predictive values are low in any one test.

Fetal hydrops

Fetal hydrops is typically diagnosed by ultrasound examination.

Treatment

Reproductive Issues

Although there is often no effective treatment to restore fertility, assisted reproductive technology with or without oocyte/sperm donation may be helpful. Prenatal genetic diagnosis may also be performed to identify whether an expanded myotonic dystrophy allele has been passed along to the embryo.

Pregnancy Complications

Due to the increased incidence of complications during pregnancy with a child with congenital myotonic dystrophy DM1, intensive obstetric and perinatal care is recommended.

Neonatal complications

- Polyhydramnios: Amniotic fluid volume reduction may be considered only if there is preterm labor or significant maternal discomfort. Methods for reducing excessive amniotic fluid volume include:
 - Amnioreduction: Amniotic fluid is suctioned to reduce the edema seen. While amnioreduction can be repeated if severe polyhydramnios recurs, this exposes the fetus to the risks of serial invasive procedures and should be done only where symptoms warrant.
 - Maternal administration of prostaglandin synthetase inhibitors. These agents stimulate fetal secretion of arginine vasopressin, which reduces renal blood flow and fetal urine flow. This has



been seen to impair production and/or enhance reabsorption of lung liquid. Fetal and maternal side effects of these drugs include constriction of the ductus arteriosus, esophageal reflux, gastritis, and emesis, which must be monitored.

- Fetal hydrops: During pregnancy, treatment of hydrops is limited. Management of hydrops in newborn babies may include:
 - Support for respiratory distress using supplemental oxygen or mechanical ventilation
 - Removal of excessive fluid from spaces around the lungs and abdomen
 - Medications to help the kidneys remove excess fluid