



**MYOTONIC
DYSTROPHY
FOUNDATION**

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Multisystemic Characteristics of DM: Reproductive and Endocrine Systems

Symptoms

Reproductive Issues

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;
 - o Elevated FSH levels can result in high estradiol:testosterone ratios, leading to gynecomastia (breast enlargement in males)

Female Infertility: Reduced fertility is seen in females with DM. Symptoms include:

- increased spontaneous abortion and stillbirth rate
- early menopause in rare cases
- little evidence of gonadal dysfunction or hypogonadism

Pregnancy Complications: Maternal complications during pregnancy may include:

- prolonged labour and delivery related to uterine dysfunction, maternal weakness, and lack of voluntary assistance
- uterine overdistention, related to polyhydramnios, which can cause preterm labour, 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 barbituates
- post-partum haemorrhage due to inadequate uterine contractions (atonic uterus) or retained placenta

Neonatal Complications: Fetal and neonatal complications in newborns with congenital DM1 may include:

- polyhydramnios (excessive accumulation of amniotic fluid due to decreased fetal swallowing) which is associated with increased risks of adverse pregnancy outcome.
 - o umbilical cord prolapse or placental abruption
 - o fetal malposition due to greater fetal mobility
 - o pre-term labor
- hydrops fetalis (severe edema in the fetus)
- fetal akinesia (reduced fetal movement)



Symptoms continued

Endocrinopathy

Insulin Resistance: In DM patients, insulin-stimulated uptake of glucose (by organs such as muscle, fat or liver) is reduced due to insulin receptor deficiencies. To compensate for this suppressed responsiveness (or insulin resistance), insulin secretion may be increased. Elevated levels of circulating insulin, increased serum glucose, and dyslipidaemia may be present. Although diabetic symptoms may be seen, the insulin resistance issues tend to be mild and rarely result in full diabetes in DM1. The prevalence of diabetes is greater in DM2.

Frontal Balding: Premature male-pattern frontal balding is seen in both DM1 and DM2.

Diagnosis

Reproductive Issues: Diagnosis of fertility issues of individuals with DM may include:

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

Insulin Resistance: Diagnosis of insulin resistance in individuals with DM typically involves blood tests that measure:

- fasting serum insulin levels
- fasting serum glucose concentration
- fasting serum glycosylated hemoglobin concentration

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's 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 DM allele has been passed along to the embryo.

Insulin Resistance: Insulin resistance can be managed in two ways.

- Life-style changes. The need for insulin can be reduced by modifying lifestyle (e.g. exercise, balanced diet, removal of majority of sugar from the diet).
- Medications. Blood glucose and insulin levels can be normalized by drugs that either prevent the liver from releasing glucose into the blood or increase the sensitivity of muscle and fat cells to insulin.



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Treatment continued

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

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 can be 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