



Endocrine System

Symptoms

Insulin Resistance

In myotonic dystrophy patients, insulin-stimulated uptake of glucose is reduced due to insulin receptor deficiencies. To compensate for suppressed responsiveness (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

Insulin Resistance

Diagnosis of insulin resistance in individuals with myotonic dystrophy typically involves blood tests that measure:

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

Treatment

Insulin Resistance

Insulin resistance can be managed in the following ways:

- Lifestyle changes: The need for insulin can be reduced by modifying lifestyle (eg. 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.

Immune System

Myotonic dystrophy is associated with a modest reduction in the amount of immunoglobulin in the blood (hypogammaglobulinemia). The production of antibodies is normal, however the antibodies do not last as long in the circulation, hence the amount in the blood at any time is somewhat reduced. The myotonic dystrophy-associated reduction of immunoglobulin appears to be well tolerated. So far there is no clear evidence that alteration is associated with an increased frequency of infection.



Tumors

People with myotonic dystrophy DM1 have an increased frequency of pilomatrixoma, a type of benign skin tumor. This type of tumor is rare in the general population but fairly common in people with myotonic dystrophy DM1. (No association between pilomatrixomas and DM2 has been reported).

Pilomatrixomas often occur around the head or neck and feel like firm lumps just beneath the surface of the skin. These tumors are benign and can be cured by surgical removal. Some researchers have suggested that DM1 may also be associated with an increased frequency of other types of tumors, such as tumors of the parathyroid, pituitary, or thymus glands. However, at this point there is no clear evidence to support this idea.