



Friedreich's Ataxia Presenting with Diabetes Mellitus in an Adolescent

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Introduction: Friedreich's ataxia (FA) is an autosomal recessive neurodegenerative disorder characterized by progressive ataxia with limb muscle weakness, absent lower limb reflexes, extensor plantar responses, dysarthria, decreased vibratory sense and proprioception. The most common molecular abnormality is a GAA trinucleotide repeat expansion in intron 1 of the *frataxin (FXN)* gene.

- Patients with FA are at risk of getting increased blood sugar levels, or glucose intolerance, and around 20% progress to overt diabetes mellitus (DM). The cause of diabetes in FA is poorly understood. Glucose intolerance and diabetes can result from a shortage in insulin secretion by the insulin-producing β cells in the pancreas, from insulin resistance, or from a combination of both.

Case

- A 15.5-year-old girl
- Weight loss, polyuria and polydipsia of 1-month duration
- Gait disturbance and pain in her soles for the last 3-years
- Neurological examination demonstrated ataxic gait pattern, pes-cavus, intentional tremor, lower limb areflexia and fleksor plantar response.
- Thyroid gland was soft, and bilaterally 4 cm on palpation

Clinical Course

- Basal and bolus insulin treatment had been initiated.
- Echocardiography were normal.
- Methimazole was added to treatment.
- *FTX* gene analysis:
Homozygous >66 GAA tri-nucleotide repeats in intron 1

Table.1: Laboratory Findings for Diabetes

| | Value | References |
|-----------------------------|----------|------------|
| Glucose (mg/dl) | 390 | |
| Insulin (μ IU/ml) | 1.53 | |
| C-peptide (ng/ml) | 1.08 | 1.1-5 |
| HbA1c | 13.4% | |
| Keton | +++ | |
| Diabetes* Autoantibodies | Negative | |

*Anti-insulin, GAD, and islet cell antibodies

Table.2: Laboratory Findings for Hyperthyroidism

| | Value | References |
|----------------------------|----------|------------|
| TSH (μ IU/ml) | 0.03 | 0.34-5.6 |
| f-T4 (n g/dl) | 1.79 | 0.61-1.12 |
| f-T3 (pg/ml) | 4.57 | 2.5-3.9 |
| Thyroid Autoantibodies* | Negative | |

* TSH receptor autoantibodies, anti thyroid peroxidase and anti thyroglobulin

Conclusions

- ✓ Friedreich Ataxia should be considered in patients who presented with diabetes and ataxia. Non-autoimmune hyperthyroidism detected in this patient is a novel finding in FA.

