

Table 1. Developmental history, clinical history and physical examination findings in the three cases

| | Features | Case 1 | Case 2 | Case 3 |
|-----------------------|-----------------------------|---|---|--|
| Development | Gender | Female | Male | Male |
| | Current age | 21 years | 25 years | 32 years |
| | Parents | Reportedly unrelated | Not related | First cousin consanguineous |
| | Birth weight | 1520 g (-4.4 SDS) | 2450 g (-3.2 SDS) | 1800 g (-2.8 SDS) |
| | Birth length | 39 cm (-5.5 SDS) | NR | 45 cm (-3.0 SDS) |
| | Birth OFC | 31 cm (-2.4 SDS) | NR | 33 cm (-1.2 SDS) |
| | Psychomotor development | Normal | Normal | Delayed |
| | Linear growth | Severe growth failure. Adult height 120 cm (-6.6 SDS) | Severe growth failure. Adult height 127 cm (-7.2SDS) | Severe growth failure. Adult height 138 cm (-5.8 SDS) |
| Clinical observations | Insulin resistance | Insulin resistance which progressed to Type 2 diabetes | Insulin resistance with reactive hypoglycaemia | Insulin resistance which progressed to Type 2 diabetes |
| | Hypertension | Present, treated | NR | Absent |
| | Hyperlipidaemia | Diagnosed at 11 years | Diagnosed at 22 years | Diagnosed at 22 years |
| | Ophthalmological assessment | Astigmatism | NR | Mild non-proliferative diabetic retinopathy |
| | Pubertal development | Tanner B2 at 9.8 years, menarche at 15.3 years | Tanner G2 at 11 years; G3 (testes 8 ml) at 13.5 years | Absent (G1 at 21 years), gynaecomastia |
| | Muscle cramps | Onset aged 2 years | Onset aged 13 years | Onset aged 22 years |
| | Alopecia | Present | Present | Present |
| | Centripetal obesity | Absent (waist circumference 72 cm) | Present | Present |
| | Acanthosis Nigricans | Present from 10.1 years | Present from 13.5 years | Present from 21 years |
| | Hypotonia | NR | NR | Present |
| | High pitched voice | Present | Present | Absent |
| | Adult gonadal status | Partial ovarian failure (due to IR?) | NR | Borderline low plasma testosterone |
| Laboratory | Insulin | Increased | Increased | Increased |
| | Creatine Kinase | Increased | Increased | Increased |
| Additional findings | Empty sella turcica | NR | NR | Present |
| | Diffuse fatty liver | Present | Present | Present |
| | Kidney anatomy | Normal kidney ultrasonography | NR | Left ectopic kidney |
| | Electromyography (EMG) | Reduced recruitment of MUAPs firing at increased frequency with increased amplitude, polyphasic potentials. Spontaneous fasciculations. | NR | Rare fibrillations and positive sharp waves. Normal MUAPs, morphology and recruitments. Muscular cramps induced by leg exercise accompanied by fasciculation |
| | Colonoscopy | NR | NR | Transverse colon polyp, no dysplasia or malignancy |

Abbreviations: MUAPs, Motor Unit Action Potentials; NR, Not Reported; OFC, occipitofrontal circumference; SDS, standard deviation score