**Supplemental Table 2.** Characteristics of non-CAH PAI patients with unidentified molecular etiology

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient** | **Age of diagnosis**  **years** | **GC deficiency** | **MC deficiency** | **Sex steroid deficiency** | | **Elevated TSH** | **Additional**  **features** | **IUGR / Short stature** |
| **Gonadal** | **Adrenal** |
| **P1** | 8.67 | + | + | \* | + | - | Yes1 | -/- |
| **P2** | 0.02 | + | + | - | + | + | Yes2 | -/- |
| **P3** | 0.01 | + | - | \* | + | - | No | -/- |
| **P4** | 16.51 | + | + | - | + | + | Yes3 | -/- |
| **P5** | 23.76 | + | - | - | + | + | Yes4 | +/+ |
| **P6** | 0.02 | + | + | - | + | + | No | +/+ |
| **P7** | 13.35 | + | - | - | + | - | Yes5 | -/- |
| **P8** | 0.11 | + | + | + | - | + | Yes6 | +/- |
| **P9** | 1.10 | + | - | \* | + | - | Yes7 | -/- |
| **P10** | 0.33 | + | + | - | + | - | No | -/- |
| **P11** | 9.09 | + | + | - | + | - | No | -/- |
| **P12** | 10.83 | + | - | - | + | - | Yes8 | -/- |

Abbreviations: PAI: Pimary adrenal insufficiency, GC: Glucocorticoid, MC: Mineralocorticoid, TSH: Thyroid stimulating hormone, IUGR: Intrauterine growth retardation.

\*The patient could not be evaluated as he/she was not in mini puberty or puberty.

1Hypogonadotropic hypogonadism, ataxia 2Nephrotic syndrome, congenital neutropenia, 3Hashimoto thyroiditis, 4Hypoplastic left ventricle, left renal agenesis, left cerebral hypoplasia, microcephaly, mental retardation, 5Eczema, prurigo nodularis, high serum IgE, mild tricuspid and mitral valve regurgitation, 6Hypospadias, cryptorchidism, 7Mature cystic teratoma in testis, low Anti Mullerian hormone (AMH), another sibling died with similar findings, 8High serum anti glutamic acid decarboxylase (anti-GAD) antibody.