**Supplemental Table 3.** Comparison of clinical and biochemical characteristics of PAI patients with identified and unidentified etiology.

|  |  |  |  |
| --- | --- | --- | --- |
|  | **PAI identified (n=29)** | **PAI unidentified (n=12)** | ***p*** |
| **Age of presentation (years)** | 1.3 ± 0.3 | 5.7 ± 2.1 | ***0.005\*\**** |
| **Consanguinity of parents (n)** | 23 | 3 | ***0.001\*\**** |
| **MC deficiency (n)** | 17 | 7 | 0.98 |
| **Sex steroid deficiency (n)** |  |  |  |
| Gonadal | 9 | 1 | 0.12 |
| Adrenal | 28 | 11 | 0.5 |
| **Karyotype (n)** |  |  |  |
| 46,XX | 10 | 2 | 0.25 |
| 46,XY | 19 | 10 |
| **Gender (n)** |  |  |  |
| Female | 15 | 2 | ***0.03\**** |
| Male | 14 | 10 |
| **ACTH (N=10-60 pg/mL)** | 1132 ± 55.14 | 992.7 ± 204.8 | 0.38 |
| **Cortisol (µg/dL)** | 0.8 ± 0.3 | 0.4 ± 0.1 | ***0.002\*\**** |
| **Cortisone (ng/mL)** | 3.8 ± 1.6 | 24.2 ± 5.1 | ***0.0001\*\*\**** |
| **Corticosterone (ng/mL)** | 0.20 ± 0.06 | 1.38 ± 0.40 | ***<0.0001\*\*\*\**** |
| **11-Deoxycorticosterone (ng/mL)** | 0.02 ± 0.007 | 0.10 ± 0.05 | ***0.02\**** |

Abbreviations: PAI: Pimary adrenal insufficiency, MC: Mineralocorticoid, ACTH: Adrenocorticotropic hormone

To convert ng/mL to nmol/L, multiply by 2.89 for corticosterone; 3.03 for 11-deoxycorticosterone; 2.75 for cortisol; 2.77 for cortisone.