Supplementary table 1 Characteristics of infants and young children with CAH, PAI and Xp21 contiguous gene deletion syndrome

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| No | Sex | Age at onset | Chief complaint | Clinical specific sign | K  (mmol/L) | Na  (mmol/L) | Diagnosis | Gene | Mutation |
| 1 | Male | 9 d | Poor feeding | None | 7.4 | 128.5 | 21-OHD CAH | CYP21A2 | c.713T>A  c.844G>T  c.955C>T |
| 2 | Female | 19 d | Fever | Hyperpigmentation  Virilization of external genitalia | 8.0 | 122 | 21-OHD CAH | CYP21A2 | c.332-339delGAGACTAC  c.710T>A c,713T>A c.844G>T c.1069C>T |
| 3 | Female | 1 m | Poor weight gain  Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 5.9 | 124.5 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.955C>T  c.1069C>T |
| 4 | Male | 2 m | Poor feeding  Poor weight gain | Hyperpigmentation | 5.6 | 120.5 | 21-OHD CAH | CYP21A2 | 293-13C>G  Rearrangement of 1-7 exon |
| 5 | Male | 15 d | Poor feeding  Poor weight gain | Hyperpigmentation | 7.8 | 118.3 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.1069C>T |
| 6 | Male | 1.8 m | Vomiting  Poor weight gain | Hyperpigmentation | 7.7 | 121 | 21-OHD CAH | CYP21A2 | Hom in c.293-13C>G |
| 7 | Male | 2 m | Vomiting  Poor weight gain | Hyperpigmentation | 7.0 | 103.5 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.844G>T  c.955C>T  c.1069C>T |
| 8 | Male | 2.5 m | Vomiting  Diarrhea  Poor weight gain | Hyperpigmentation | 6.1 | 115 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.518T>A  c.710T>A  c.713T>A  c.719T>A  c.1069C>CT  c.844G>T |
| 9 | Female | 10 d | Poor weight gain  Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 6.4 | 111.2 | 21-OHD CAH | CYP21A2 | c.713T>A  c.719T>A  c.1069C>T  c.651+37A>G |
| 11 | Male | 1.8 m | Vomting  Diarrhea  Poor weight gain | Hyperpigmentation | 7.6 | 105.5 | 21-OHD CAH | CYP21A2 | Hom in c.293-13C>G  Exon 1,3 large deletion |
| 12 | Female | 1.4 m | Poor weight gain  Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 7.4 | 110 | 21-OHD CAH | NA | NA |
| 13 | Female | 19 d | Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 6.4 | 129.6 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.518T>A |
| 14 | Male | 22 d | Vomting  Poor weight gain | Virilization of external genitalia | 10.7 | 104.1 | 21-OHD CAH | NA | NA |
| 15 | Male | 20 d | Poor weight gain | Hyperpigmentation | 7.2 | 117.9 | 21-OHD CAH | NA | NA |
| 16 | Male | 17 d | Poor weight gain | Hyperpigmentation | 8.8 | 109.3 | 21-OHD CAH | NA | NA |
| 17 | Male | 28 d | Poor weight gain  Vomiting | Hyperpigmentation | 5.86 | 105.2 | 21-OHD CAH | CYP21A2 | c.293-13C>G  Exon 1-7 large deletion |
| 18 | Female | 11 d | Fever  Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 6.5 | 128.6 | 21-OHD CAH | CYP21A2 | Hom in c.293-13C>G |
| 19 | Female | 13 d | Vomiting  Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 8.9 | 110.4 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.923dupT  Exon 1,3,4,6,7 large deletion |
| 20 | Male | 15 d | Vomiting | Hyperpigmentation | 6.4 | 127.9 | 21-OHD CAH | NA | NA |
| 21 | Female | 4 m | Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 5.6 | 132.1 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.518T>A |
| 22 | Female | 4 m | Virilization of external genitalia | Hyperpigmentation  Virilization of external genitalia | 5.3 | 133.5 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.518T>A |
| 23 | Male | 28 d | High 17-OH progesterone in newborn screening | Hyperpigmentation | 6.3 | 120 | 21-OHD CAH | CYP21A2 | c.293-13C>G  c.1451G>C  c.1455delG |
| 24 | Female | 1 m | Virilization of external genitalia  Diarrhea | Hyperpigmentation  Virilization of external genitalia | 6.4 | 121 | 21-OHD CAH | NA | NA |
| 25 | Male | 21 d | Fever  Vomiting | Hyperpigmentation | 6.5 | 94.7 | 21-OHD CAH | CYP21A2 | Hom in c.293-13C>G |
| 26 | Male | 3 m | Vomiting  Poor weight gain | Hyperpigmentation | 6.3 | 122.5 | 21-OHD CAH | NA | NA |

PE: physical examination; d: days; m: months; y: years; FC: fludrocortisone; HC: hydrocortisone; CAH: congenital adrenal hyperplasia;

PAI: primary adrenal insufficiency; 21-hydroxylase deficiency: 21-OHD; hom: homozygous; NA: not available.