

Supplementary Material

Second-tier screening results of 21-OHD CAH patients diagnosed by neonatal screening

Supplementary Table 1a. Screening results of SW-21-OHD patients diagnosed by neonatal screening							
Number	17-OHP (1 st tier)	LC-MS/MS 17-OHP	LC-MS/MS 11-deoxycortisol	LC-MS/MS 21-deoxycortisol	LC-MS/MS androstenedione	LC-MS/MS cortisol	(21-S + 17- OHP)/F
1	56,5	45,782	0,318	12,808	1,482	1,65	35,51
2	67	44,297	1,107	2,051	3,022	2,711	17,10
3	36,1	53,554	0,582	59,765	4,34	17,825	6,36
4	47,09	56,576	2,282	31,177	4,068	8,59	10,22
5*	48,6	10,2	1,73	0,2	2,95	69,19	0,14
6	49,08	14,913	0,16	10,12	3,689	2,943	8,51
7*	51,07	23,326	0,225	11,798	3,488	48,426	0,47
8	54,93	51,979	0,984	46,131	5,871	14,389	6,82
9	57,15	26,46	0,738	7,291	10,105	11,278	2,99
10	58,15	125,232	0,729	9,345	15,99	6,775	19,86
11	58,61	24,312	1,269	3,624	5,595	5,755	4,85
12	60,8	262,18	4,828	12,507	56,834	12,093	22,71
13	60,8	156,786	3,049	23,848	54,2	8,293	21,78
14	60,8	236,273	2,325	11,072	34,75	3,267	75,71
15	67	44,297	1,107	2,051	3,022	2,711	17,10
16	67	42,87	0,91	2,054	5,049	5,938	7,57
17	67	129,071	4,086	2,986	72,61	13,565	9,74
18	67	289,375	3,17	34,093	65,5	37,462	8,63
19	67,1	428,274	5,465	14,074	81,95	11,304	39,13
20	67,1	202,755	0,338	45,392	7,546	7,339	33,81
21	67,1	133,151	3,001	3,599	19,63	3,88	35,24
22	67,1	48,639	1,721	22,993	3,576	11,195	6,40
23	67,1	94,287	0,454	10,623	3,051	4,355	24,09
24	67,1	415,506	10,118	30,782	155,89	52,333	8,53
25	67,1	146,328	1,803	31,862	24,48	13,103	13,60
26	67,1	124,131	1,425	18,07	13,017	3,155	45,07
27	68	1686,86	7,579	49,833	132,937	20,2	85,97
28	68	272,769	8,522	5,114	72,693	12,612	22,03
29	68	127,797	1,639	20,378	17,371	5,178	28,62
30	68	165,82	0,873	21,539	16,121	4,783	39,17
31	68	162,558	2,661	15,923	29,655	7,578	23,55
32	68	36,963	1,608	6,638	13,16	4,545	9,59
33	68	25,566	0,647	59,52	2,597	25,662	3,32
34	68	27,576	0,704	14,438	3,162	11,723	3,58
35	68	343,933	7,167	12,064	98,05	11,266	31,60
36	68	53,576	0,586	14,488	6,603	5,091	13,37
37	68	19,417	0,425	3,364	3,064	0,599	38,03
38	70,1	705,13	8,137	15,088	175,458	8,744	82,37
39	70,1	30,529	0,687	16,434	3,002	4,262	11,02
40	70,1	693,908	1,491	33,123	37,402	7,955	91,39
41	70,13	61,969	2,25	16,023	19,373	23,17	3,37
42	70,13	91,355	1,99	29,691	13,76	5,059	23,93
43	70,13	151,221	1,395	10,396	23,143	13,339	12,12
44	70,13	94,764	1,778	8,955	6,746	4,258	24,36
45	70,13	967,571	4,175	23,647	64,903	9,708	102,10
46	70,13	50,049	0,796	16,053	19,507	3,578	18,47
47	76,7	394,795	2,884	165,655	8,827	26,081	21,49
48	76,74	58,225	4,15	37,687	11,6	15,257	6,29
49	77	72,652	3,912	16,629	5,641	2,097	42,58
50	79,6	160,071	2,238	6,564	32,58	5,809	28,69
51	80	18,86	0,904	4,86	1,595	2,339	10,14
52	81,8	326,502	3,591	26,023	47,841	11,424	30,86
53	81,8	249,379	0,954	55,351	4,332	6,324	48,19
54	84,4	165,983	0,657	4,605	35,179	7,534	22,64
55	84,4	88,312	0,922	30,525	9,164	4,103	28,96
56	85,9	1259,83	9,579	12,604	106,4	8,969	141,87
57	85,9	58,812	2,093	4,427	23,11	5,039	12,55
58	85,9	160,446	1,148	129,735	65,36	18,867	15,38
59	86	91,658	0,501	21,653	11,88	4,635	24,45
60	67,000	289,000	3,170	34,090	65,500	37,462	8,6
61	63,23	29,055	0,573	7,796	6,068	2,674	13,781
62	76,72	87,08	1,29	11,75	9,37	9,00	10,90
63	137.30	263.345	7.177	40.656	90.44	29.957	10,14
64	44.29	19.66	0.62	0.052	1.53	1.499	14,28
65	96.00	908.457	27.84	0.492	346.9	41.275	22,14
66	96.00	58.734	3.847	0.020	6.832	12.696	4,83
67	137.30	262,59	0.83	37	23,1	9	32,47
68	90	168,37	0,86	24,56	34,28	6,38	30,23
69	95	287,35	9,32	0,84	43,77	6,8	42,38
70	90	322,55	1,37	41,99	20,9	10,73	33,97
71	95	73,6	1,91	22,94	9,19	9,04	10,67
72	90	234,36	5,56	9,14	44,97	9,04	26,93
73	90.00	321.00	1.37	41.90	20.90	10.70	36.29
74	90.00	104.40	0.28	27.23	7.70	5.44	24.19
75	90.00	220.00	1.18	28.93	9.15	8.10	30.73
76	46.60	41.47	0.25	43.21	3.98	2.33	36.34
77	95.00	73.60	1.91	22.94	9.19	9.04	10.67
*Sampled after hydrocortisone treatment, SW-21-OHD: salt-wasting 21-alpha hydroxylase deficiency, 17-OHP: 17α-hydroxyprogesterone, LC-MS/MS: liquid chromatography-tandem mass spectrometry							

Supplementary Table 1b. Screening results of SV-21-OHD patients diagnosed by neonatal screening.							
Number	17-OHP (1 st tier)	LC-MS/MS 17-OHP	LC-MS/MS 11-deoxycortisol	LC-MS/MS 21-deoxycortisol	LC-MS/MS androstenedione	LC-MS/MS cortisol	(21-S + 17- OHP)/F
1	24,22	24,467	0,579	29,446	0,776	14,979	3,60
2	15,32	6,634	0,699	3,822	1,52	4,44	2,35
3	18,71	10,346	0,627	1,543	0,975	7,897	1,51
4	19,83	5,664	0,217	0,307	0,301	1,953	3,06
5	23,96	10,475	0,517	1,717	0,789	3,174	3,84
6	26,66	22,967	0,901	4,23	1,152	46,393	0,59
7	27,55	10,47	1,921	2,001	5,482	52,978	0,24
8	31,45	7,676	0,188	0,95	0,253	1,559	5,53
9	31,74	12,257	1,415	2,986	0,758	26,116	0,58
10	54,98	70,357	4,214	0,001	8,338	63,87	1,10
11	33,81	10,87	0,618	0,604	3,439	2,61	4,76
12	44,29	19,66	0,62	0,052	1,53	14,99	1,33
13	24,22	24,467	0,579	29,446	0,776	14,979	3,60
14	60,09	14,145	4,443	0,06	0,66	8,325	1,71

SV-21-OHD: simple virilizing 21-alpha hydroxylase deficiency, 17-OHP: 17 α -hydroxyprogesterone, LC-MS/MS: liquid chromatography-tandem mass spectrometry

Supplementary Table 1c. Screening results of NC-21-OHD patients diagnosed by neonatal screening							
Number	17-OHP (1 st tier)	LC-MS/MS 17-OHP	LC-MS/MS 11-deoxycortisol	LC-MS/MS 21-deoxycortisol	LC-MS/MS androstenedione	LC-MS/MS cortisol	(21-S + 17- OHP)/F
1	9,49	2,526	0,655	1,696	0,492	32,621	0,13
2	9,9	7,864	0,406	3,393	0,741	31,551	0,36
3	10	3,64	0,393	0,01	0,255	2,073	1,76
4	10	4,17	0,669	1,906	1,253	48,39	0,13
5	10,84	4,416	0,575	3,356	0,62	35,635	0,22
6	11,54	3,326	0,439	4,615	0,637	45,508	0,17
7	11,56	9,197	0,052	4,548	0,569	44,156	0,31
8	12,31	5,014	0,109	2,673	0,557	39,882	0,19
9	14,92	2,04	0,9	0,314	1,1	1,998	1,18
10	15	3,644	0,372	3,142	0,724	103,175	0,07
11	16,71	5,289	0,805	0,175	0,493	1,058	5,16
12	20,87	11,491	1,231	1,684	1,856	33,811	0,39
13	22,82	7,251	0,525	5,754	0,478	18,99	0,68
14	31,19	15	5,418	0,137	0,801	15	1,01
15	11,59	3,891	0,603	6,439	0,461	6,208	1,66
16	12,16	8,997	0,115	3,679	0,447	5,742	2,21
17	24,93	6,767	0,264	2,208	6,093	25,353	0,35
18	26,74	7,137	0,944	2,636	1,155	4,447	2,20
19	28,22	11,646	2,968	0,19	3,067	7,247	1,63
20	30,53	12,115	0,523	4,889	1,135	9,827	1,73
21	40,23	10,852	0,717	0,799	0,924	5,848	1,99
22	9,49	2,526	0,655	1,696	0,492	32,621	0,13

NC-21-OHD: neonatal screening 21-alpha hydroxylase deficiency, 17-OHP: 17 α -hydroxyprogesterone, LC-MS/MS: liquid chromatography-tandem mass spectrometry