

Supplement 1: Detailed genotypes

	sex	allele 1	allele 2	genetic group	clinical form
1	m	DelCYP21B	DelCYP21B	0	SW
2	m	DelCYP21B	DelCYP21B	0	SW
3	m	DelCYP21B	DelCYP21B	0	SW
4	m	DelCYP21B	DelCYP21B	0	SW
5	m	DelCYP21B	DelCYP21B	0	SW
6	m	Exon 8 Gln318t	DelCYP21B	0	SW
7	m	DelCYP21B	DelCYP21B	0	SW
8	m	DelCYP21B	DelCYP21B	0	SW
9	m	DelCYP21B	DelCYP21B	0	SW
10	m	DelCYP21B	DelCYP21B	0	SW
11	m	Exon 6 Ile236Asn + Val237Glu + Met239Lys	DelCYP21B	0	SW
12	m	DelCYP21B	DelCYP21B	0	SW
13	m	DelCYP21B	DelCYP21B	0	SW
14	m	Exon 8 Gln318t	Exon 8 Gln318t	0	SW
15	m	DelCYP21B	DelCYP21B	0	SW
16	m	DelCYP21B	DelCYP21B	0	SW
17	m	Exon 4 Ile172Asn + Exon 6 Ile236Asn, Val237Glu, Met239 Lys + Exon 7 Val281Leu	Exon 3 del (8bp)	0	SV
18	m	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
19	m	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
20	m	Intron 2 PM 656 A/C→G	Intron 2 PM 656 A/C→G	A	SW
21	m	Intron 2 PM 656 A/C→G	Exon 3 del (8bp) + Exon 8 Arg356Trp	A	SW
22	m	Intron 2 PM 656 A/C→G	Exon 8 Arg316X	A	SW
23	m	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
24	m	Intron 2 PM 656 A/C→G	Intron 2 PM 656 A/C→G	A	SW
25	m	Intron 2 + Exon 1+3 (In2: PM 656 A/C→G; Ex1: Pro30Leu; Ex3: del 8bp)	Intron 2 PM 656 A/C→G	A	SW
26	m	Intron 2 PM 656 A/C→G	Intron 2 PM 656 A/C→G	A	SW
27	m	Intron 2 PM 656 A/C→G	Exon 4+6+7 (4:Ile172Asn; 6: Ile236Asn, Val237Glu, Met239 Lys; 7: Val281Leu+PM Nukleotid 1761 insert T)	A	SW
28	m	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
29	m	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
30	m	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
31	m	Intron 2 PM 656 A/C→G	Exon 10 Arg483Pro	A	SW
32	m	Intron 2 PM 656 A/C→G	Intron 2 PM 656 A/C→G	A	SW
33	m	Intron 2 PM 656 A/C→G	Exon 3 Arg105Pro	A	SW
34	m	Intron 2 PM 656 C→G	Intron 2 PM 656 C→G	A	SV
35	m	Exon 7+8 (7: Val281Leu+insert T Nukleotid 1761; 8: Gln318t+Arg356Trp)	Exon 4 PM Ile172Asn	B	SW
36	m	In 2 + Ex 8 (In 2: PM 656 A/C→G; Ex 8: Arg356Trp)	Exon 4 PM Ile172Asn	B	SW
37	m	Exon 4 PM Ile172Asn	Exon 7 PM Val281Leu	B	SV
38	m	In 2+Ex 3 (In2: PM 656 A/C→G; Ex3: del 8bp)	Exon 4 PM Ile172Asn	B	SV
39	m	Exon 4 Ile172Asn	Exon 8 Gln318t	B	SV

40	m	Exon 4 Ile172Asn	Exon 3 del (8bp)	B	SV
		Exon 6 Iso236Asn,Val237Gln,Met239 Lys + Exon 7 Val281Leu			
41	m		Exon 4 PM Ile172Asn	B	SV
42	m	Intron 2 PM 656 A/C→G	Exon 4 PM Ile172Asn	B	SV
43	m	no mutation found	no mutation found	D	SW
44	m	no mutation found	no mutation found	D	SW
45	m	no mutation found	no mutation found	D	SW
46	m	Intron 2 PM 656 A/C→G	no mutation found	D	SV
47	w	DelCYP21B	DelCYP21B	0	SW
48	w	DelCYP21B	DelCYP21B	0	SW
49	w	Exon 8 Arg356Trp	DelCYP21B	0	SW
50	w	DelCYP21B	DelCYP21B	0	SW
51	w	DelCYP21B	DelCYP21B	0	SW
52	w	DelCYP21B	DelCYP21B	0	SW
53	w	DelCYP21B	DelCyp21B	0	SW
54	w	DelCYP21B	DelCYP21B	0	SW
55	w	Exon 3 del (8bp)	Exon 8 Gln318t	0	SW
56	w	Exon8 Gln318t	DelCYP21B	0	SW
57	w	DelCYP21B	DelCYP21B	0	SW
58	w	DelCYP21B	Exon 8 Gln318t	0	SW
59	w	DelCYP21B	DelCYP21B	0	SW
60	w	Exon 3 del (8bp)	DelCYP21B	0	SW
61	w	Exon 3 del (8bp)	DelCYP21B	0	SW
62	w	DelCYP21B	DelCYP21B	0	SW
63	w	DelCYP21B	DelCYP21B	0	SW
64	w	DelCYP21B	DelCYP21B	0	SW
65	w	Exon 8 Gln318t+Arg356Trp	Exon 7 Val281Leu + Exon 8 Gln318t+Arg356Trp	0	SW
66	w	Exon 8 Gln318t	Exon 8 Gln318t	0	SW
67	w	Exon 7 Val281Leu + Exon 8 Arg356Trp	Exon 3 del (8bp)	0	SW
68	w	Exon 4 Ile172Asn + Exon 6 Ile136Asn,Val237Glu,Met239 Lys + Exon 7 Val281Leu	DelCYP21B	0	SV
69	w	Intron 2 PM 656 A/C→G	Exon 3 del (8bp)	A	SW
70	w	Intron 2 PM 656 A/C→G	Exon 8 PM Gln318t	A	SW
71	w	Intron 2 PM 656 A/C→G; Exon 7 PM Val281Leu	DelCYP21B	A	SW
72	w	Intron 2 PM 656 A/C→G; Exon 7 PM Val281Leu	DelCYP21B	A	SW
73	w	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
74	w	Intron 2 PM 656 A/C→G	Exon 8 Arg356Trp	A	SW
75	w	Intron 2 PM 656 A/C→G	Exon 7 1761 insert T	A	SW
76	w	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
77	w	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
78	w	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
79	w	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
80	w	Intron 2 PM 656 A/C→G	Exon 8 Arg356Trp	A	SW
81	w	Intron 2 PM 656 A/C→G	Exon 8 Gln318t	A	SW
82	w	Intron 2 PM 656 A/C→G	Exon 8 Gln318t	A	SW
83	w	Intron 2 PM 656 A/C→G	Exon 3 del (8bp)	A	SW
84	w	Intron 2 PM 656 A/C→G	Exon 3 del (8bp)	A	SW
85	w	Intron 2 PM 656 A/C→G	In 2 +Ex 1+3 (In 2:PM 656 A/C→G; Ex1: Pro30Leu; Ex3: del 8bp)	A	SW

86	w	Intron 2 PM 656 A/C→G	DelCYP21B	A	SW
87	w	Exon 4 PM Ile172Asn	Exon 4 PM Ile172Asn	B	SW
88	w	Exon 7+8 (7: Val281Leu+insert T bei Nukleotid 1761; 8: Gln318t+Arg356Trp)	Exon 4 PM Ile172Asn	B	SW
89	w	Intron 2 PM 656 A/C→G	Exon 4 PM Ile172Asn	B	SV
90	w	Intron 2 PM 656 C-->G	Exon 4 PM Ile172Asn	B	SV
91	w	Exon 4 Ile172Asn	Exon 3 del (8bp)	B	SV
92	w	Intron 2 PM 656 A/C→G	Exon 4 PM Ile172Asn	B	SV
93	w	DelCYP21B	Exon 4 PM Ile172Asn	B	SV
94	w	Exon 4 PM Ile172Asn	Exon 4 PM Ile172Asn	B	SV
95	w	Exon 4 PM Ile172Asn	Exon 4 PM Ile172Asn	B	SV
96	w	Ile172>Asn	DelCYP21B	B	SV
97	w	Exon 7 Val281Leu + Exon 8 Gln318t + Exon 3 del (8bp)	no mutation found	D	SW
98	w	no mutation found	no mutation found	D	SV

Supplement 2: DHEAS values [ng/mL] of healthy children (Ref. 25)

Boys			
age	n	median	quartiles
1 year	n=26	357	129, 667
2-4 years	n=27	66	1, 145
5-6 years	n=22	132	46, 187
7-8 years	n=28	133	50, 233
9-10 years	n=22	464	207, 865
11-12 years	n=20	860	416, 1575
13-14 years	n=27	1510	1160, 1870
15-16 years	n=31	1460	1068, 2070
17-18 years	n=24	1430	1188, 2548

Girls			
1 year	n=24	246	92, 500
2-4 years	n=19	3.6	1, 73.5
5-6 years	n=21	57	10, 163
7-8 years	n=20	343	205, 436
9-10 years	n=21	444	240, 602
11-12 years	n=28	463	260, 1248
13-14 years	n=27	1020	628, 1630
15-16 years	n=20	1610	553, 2635
17-18 years	n=18	1435	925, 2090