Rev 2024-04-25

Product Note: CAH Nomenclature Update 2024

The CAH StripAssay® (REF 4-380, 4-380-A, 4-380-TRIAL) is a qualitative genetic test for the targeted analysis of 11 common mutations in the *CYP21A2* gene causing congenital adrenal hyperplasia (CAH). The test is used as an aid to confirm the presence of *CYP21A2* mutations in patients with a suspected diagnosis of CAH and as a second-tier diagnostic test in newborn screening programs. In addition, approximately 50% of large deletions or conversions can be detected. *CYP21A1P/CYP21A2* chimeric genes with a junction site downstream cluster E6 and complete heterozygous *CYP21A2* deletions will not be detected.

The *CYP21A2* gene may harbour polymorphisms affecting the numbering of oligonucleotides and codons. Thus, differences in nomenclature exist in the literature which result from different reference sequences. ViennaLab uses the RefSeq NM_000500.9 as it is recommended by the EMQN guidelines (Baumgartner-Parzer *et al*., 2020) for genetic testing of 21-OHP deficiency. Beginning with RefSeq NM_000500.7 there was a shift of three nucleotides, thus P30L (c.89C>T) became P31L (c.92C>T) etc.

Gene name: *Steroid 21-hydroxylase*
Gene abbreviation: *CYP21A2*

legacy name			**HGVS nomenclature**		**RefSNP**
new		old	new (NM_000500.9)	old (NM_000500.6)	
P31L		(P30L)	c.92C>T	c.89C>T	rs9378251
I2 splice		I2 splice	c.293-13A/C>G	c.290-13A/C>G	rs6467
Del 8 bp E3		Del 8 bp E3	c.332_339del	c.329_336delGAGACTAC	rs387906510
I173N		(I172N)	c.518T>A	c.515T>A	rs6475
Cluster E6	I237N	(I236N)	c.707T>A	c.707T>A	rs111647200
	V238E	(V237E)	c.710T>A	c.710T>A	rs12530380
	M240K	(M239K)	c.716T>A	c.716T>A	rs6476
V282L		(V281L)	c.844G>T	c.841G>T	rs6471
L308fs		(L307fs)	c.923dup	c.920_921insT	rs267606756
Q319X		(Q318X)	c.955C>T	c.952C>T	rs7755898
R357W		(R356W)	c.1069C>T	c.1066C>T	rs7769409
P454S		(P453S)	c.1360C>T	c.1357C>T	rs6445
R484P		(R483P)	c.1451G>C	c.1448G>C	rs200005406

Twenty to 25% of pathogenic variants are large *CYP21A2* deletions, duplications or chimeric genes. Therefore, assessment of the *CYP21A2* copy number variation (CNV) is crucial for diagnosis. The combination of both, CAH StripAssay® and CAH RealFast™ CNV Assay is a faster and more cost-efficient targeted analysis compared to sequencing the whole gene and CNV analysis by MLPA. In total, ViennaLab CAH assays have a detection rate of up to 94%.

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