NEW!

Newborn screening

Make NBS efficient and accessible with BioVendor IVD assays

RealFast™ Assays and StripAssays® manufactured by ViennaLab Diagnostics are optimized and validated for use in genetic testing of Spinal Muscular Atrophy (SMA), Severe Combined Immunodeficiency (SCID), Sickle Cell Disease (SCD), as well as Thalassemia, Cystic Fibrosis (CF) and Congenital Andrenal Hyperplasia (CAH).

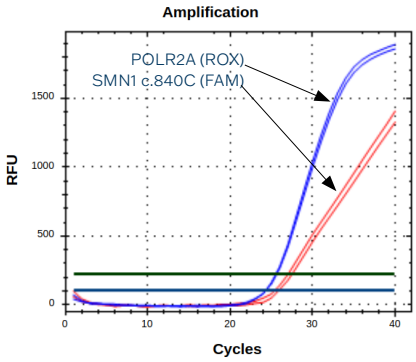
BioVendor Group®

Spinal Muscular Atrophy (SMA)

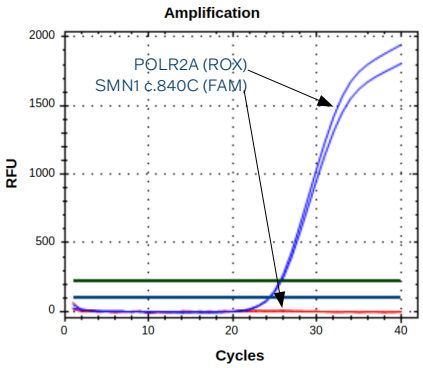
Spinal muscular atrophy (SMA) is an autosomal recessive neurodegenerative disease. SMA affects the alpha-motoneurons of the central nervous system. The homozygous *SMN1* exon 7 deletion (Δ7SMN1) is causative for >95% of SMA patients.[1]

The prevalence of SMA is approximately 1/10,000.[2]

With the **SMN1 RealFast™ Assay**, newborns can be tested for the presence or absence of SMN1 c.840C in a very short time, allowing for immediate therapeutic treatment measures.



SMN1 c.840C is present. The DNA sample used contains one or more SMN1 copies. Δ7SMN1-associated SMA can therefore be excluded. The *POLR2A* gene serves as a PCR control.

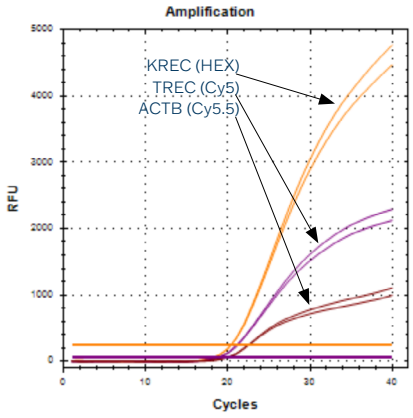


SMN1 c.840C is absent. The DNA sample used from an SMA patient has 0 copies of SMN1. The *POLR2A* gene serves exclusively as a PCR control.

Severe Combined Immunodeficiency (SCID)

Severe combined immunodeficiency (SCID) comprises a heterogeneous group of diseases characterized by a deficiency of T or B cells and in some cases also of natural killer (NK) cells.[3]

With the **SCID-XLA mpx RealFast™ Assay**, the copy number of T-cell receptor excision circles (TRECs) and kappa-deleting recombination excision circles (KRECs) can be determined in newborns rapidly.



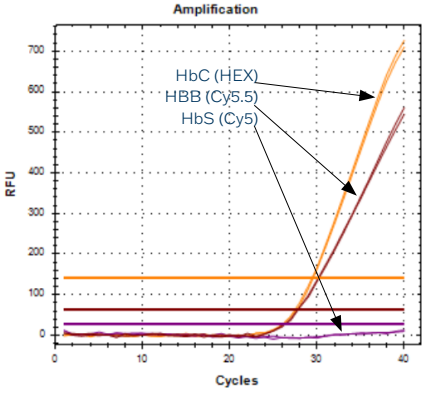
Amplification of ACTB, KREC and TREC gene copy numbers. The assay contains a plasmid which serves as a positive control and as a calibrator for the determination of ACTB, KREC and TREC gene copies in patient samples.

Sickle Cell Disease (SCD)

SCD refers to a group of inherited red blood cell disorders characterized by mutations in the β-globin gene (*HBB*).

The most common mutation in the *HBB* gene is c.20A>T, which defines the HbS allele. Another mutation in the *HBB* gene (c.19G>A) characterizes the HbC allele. Erythrocytes containing mutated β-globin subunits can polymerize, become sickle-like shaped and are prone to hemolysis. SCD is inherited as an autosomal codominant trait.[4]

With the **HbS-HbC mpx RealFast™ Assay**, newborns can be tested for HbS and HbC efficiently.



HbC heterozygous DNA sample.

Congenital Adrenal Hyperplasia (CAH)

Congenital adrenal hyperplasia (CAH) summarizes metabolic disorders that lead to inadequate synthesis of adrenal steroid hormones (cortisol, aldosterone). By far the most frequent form of CAH is caused by genetic variants in the *CYP21A2* gene encoding for steroid 21-hydroxylase.

The clinical presentation is highly variable and includes virilization of female newborns and lifethreatening salt-wasting conditions.[5]

Since early treatment is recommended, CAH newborn screening programs based on 17-hydroxyprogesteron (17-OHP) levels have been introduced in many countries. However, due to the high false-positive rate of 17-OHP assays, a second-tier test using liquid chromatography-tandem mass spectroscopy or molecular genetic analysis may be necessary.[6]

ViennaLab Diagnostics has developed two complementary genotyping assays for early and reliable confirmation of CAH:

The combination of **CAH StripAssay®** and **CAH RealFast™ CNV Assay** pinpoints the most frequent *CYP21A2* variants including deletions and duplications.

For more detailed information, please refer to the product specific CAH leaflet.

Cystic Fibrosis (CF)

Cystic Fibrosis (CF) is the most common lifelimiting autosomal recessive disorder in the Caucasian population.
The disease incidence is estimated to be 1 in 2,500 to 4,000 live births.

Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) is an anion channel which is responsible for the salt-, fluid- and pH-balance in secretory and absorptive epithelial tissues. Mutations in the *CFTR* gene lead to dysfunction of chloride transport across cell membranes.
Affected children commonly experience decreased pulmonary function along with persistent respiratory infections, pancreatic insufficiency and malnutrition.

CFTR genotyping enables early diagnosis in newborn screening and minimizes emotional stress for parents.

ViennaLab Diagnostics offers reliable and convenient reverse-hybridization and realtime PCR assays **tailored** to **population specific mutations** in **different regions**.

For more detailed information, please refer to the product specific CF leaflet.

Thalassemia

In healthy adults 97-98% of total hemoglobin (Hb) is HbA, consisting of two α-globin and two β-globin polypeptides (α2β2).
Abnormalities in the structure and synthesis of both globin chains lead to an imbalance causing the two main types of thalassemia: α-thalassemia and β-thalassemia.

Thalassemias are a major public health burden, particularly in Mediterranean countries, the Middle East, Asia, India, Iran, Arabian Peninsula and parts of Africa. For the large majority of affected individuals there is only supportive management but no ultimate cure. Health authorities therefore focus on prevention programs based on carrier and premarital screening. Since only a few α- and β-globin alleles are prevalent in each at-risk population, large-scale screening programs are feasible but require simple and automated test procedures.

ViennaLab Diagnostics offers the globally applicable **α-Globin StripAssay®**, three tailored **β-Globin StripAssays®** for the Mediterranean area (MED), India & Middle East (IME) and Southeast Asia (SEA), as well as the **β-Thal Modifier StripAssay®** and soon the **Thalassemia NGS Assay.**

For more detailed information, please refer to the product specific Thalassemia brochure.

Order Information

Category	Clinical Topic	Product Name	REF	Regulatory Status	Unit Size	Dyes
NEW!	SMA	SMN1 RealFast™ Assay	7-700	RUO	500 Rxn 100 Rxn	FAM, ROX
NEW!	SCID	SCID-XLA mpx RealFast™ Assay	7-710	RUO	500 Rxn 100 Rxn	HEX, Cy5, Cy5.5
NEW!	SCD	HbS-HbC mpx RealFast™ Assay	7-280	RUO	100 Rxn	HEX, Cy5, Cy5.5
	CAH	CAH StripAssay®	4-380	CE IVDR	20 tests	n.a.
	CAH	CAH RealFast™ CNV Assay	7-410	CE IVDR	100 Rxn	FAM, HEX
	CF	CF StripAssay®	4-410	CE IVDR	10 tests	n.a.
	CF	CF StripAssay® GER	4-430	CE IVDR	10 tests	n.a.
	CF	CF StripAssay® TUR	4-420	CE IVDR	10 tests	n.a.
	CF	CF StripAssay® EXT	4-440	CE IVDR	10 tests	n.a.
NEW!	CF	CF F508del RealFast™ Assay	7-260 7-263	CE IVDD	100 Rxn 32 Rxn	FAM, HEX
	Thalassemia	α-Globin StripAssay®	4-160	CE IVDR	10 tests	n.a.
	Thalassemia	β-Globin StripAssay® MED	4-130	CE IVDR	20 tests	n.a.
	Thalassemia	β-Globin StripAssay® IME	4-140	CE IVDR	20 tests	n.a.
	Thalassemia	β-Globin StripAssay® SEA	4-150	CE IVDR	20 tests	n.a.
	Thalassemia	β-Thal Modifier StripAssay®	4-170	CE IVDD	20 tests	n.a.

Note: The selective probe selection enables the combination of SMN1 RealFast™ Assay with SCID-XLA mpx RealFast™ Assay, as well as a combination of SMN1 RealFast™ Assay with HbS-HbC mpx RealFast™ Assay in the same run.

References:

[1] Prior et al., 2011; Rao et al., 2018; Chien et al., 2017.
[2] Reviews siehe Luo et al. 2014; Rao et al., 2018; Scheffer et al., 2001.
[3] Serana F et al. Use of V(D)J recombination excision circles to identify T- and B-cell defects and to monitor the treatment in primary and acquired immunodeficiencies. J Transl Med. 2013 May 9;11:119. doi: 10.1186/1479-5876-11-119.
[4] Kato GJ, Piel FB, Reid CD, et al. Sickle cell disease. Nat Rev Dis Primers. 2018; 4:18010.
[5] Speiser PW, White PC. Congenital adrenal hyperplasia. N Engl J, Med. 2003;349:776-88.
[6] White PC. Update on diagnosis and management of congenital adrenal hyperplasia due to 21-hydroxylase deficiency. Curr Opin Endocrinol Diabetes Obes. 2018 Jun; 25(3):178-184.

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