

Molecular Genetics - Aldolase B-Gene (ALDOB) (A149P, A174D, N334K)

Flörke / Kappert

793

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

Aldolase B: A 149 P

Aldolase B: A 174 D

Aldolase B: N 334 K

May (3)

Nov. (6)

Registration deadline:

27.02.26

07.08.26

Sample shipment:

28.04.26

06.10.26

Closing Date:

29.05.26

06.11.26

Molecular Genetics - Alpha-Globin and beta-Globin

Frömmel / Méndez

794

Samples : 2 samples of 0.04 mL
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

Mutations in alpha-globin gene (max. 4)

Genotype alpha-globin gene (nomenclature haemoglobinopathies)

Evaluation alpha-globin gene Mutations in beta globin gene (max. 4)

Genotype beta-globin gene (nomenclature haemoglobinopathies)

Evaluation beta-globin gene Report with findings from alpha- and beta- globin gene together

May (3)

Nov. (6)

Registration deadline:

27.02.26

07.08.26

Sample shipment:

28.04.26

06.10.26

Closing Date:

29.05.26

06.11.26

Molecular Genetics - Alpha1-Antitrypsin-Gene (SERPINA1) (PiM, PiS, PiZ)

Flörke / Kappert

743

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Alpha-1 antitrypsin S-Locus

Alpha-1 antitrypsin Z-Locus

May (3)

Nov. (6)

Registration deadline:

27.02.26

07.08.26

Sample shipment:

28.04.26

06.10.26

Closing Date:

29.05.26

06.11.26

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual
 (grace period: 30. May 2026)

ACE Insertion/Deletion (I/D) polymorphism

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Antithrombin (AT3; SERPINC1 Gene)

Christensen / Kappert

792

Samples : 2 Samples of 50 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual
 (grace period: 30. May 2026)

Analysis of the SERPINC1 gene for small base variations (SNVs) and/or copy number variations (CNVs)

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Apolipoprotein B APOB, R3500Q (HGVS: c.10580G>A; p.Arg3527Gln), R3500W (HGVS: c.10579C>T; p.Arg3527Trp))

Christensen / Kappert

771

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Apo B-100 (R3500Q; R3500W)

Annotation for the evaluation: The successful participation requires the correct identification if the variants R3500Q (HGVS: c.10580G>A; p.Arg3527Gln) und R3500W (HGVS: c.10579C>T; p.Arg3527Trp) are present (homozygous/heterozygous) or not (negative) in the DNA samples.

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Apo E (genotypes 2, 3, 4)

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 2 samples of 0.5 ml
 Sample properties: Whole blood
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Apo E (genotypes 2, 3, 4)

	Apr. (2)	Oct. (5)
Registration deadline:	09.01.26	03.07.26
Sample shipment:	10.03.26	01.09.26
Closing Date:	10.04.26	02.10.26

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 months / 24 months

participation according to Rili-BÄK:
 yearly

AZFa- AZFb-, AZFc- deletion

extension analysis of the deletion, interpretation and recommendations

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 month / **24 months**

participation according to Rili-BÄK:
 yearly

Analysis of the BRCA1/BRCA2 genes for small base variations (SNVs) and/or copy number variations (CNVs)

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - C-C Motif Chemokine Receptor 5

Wenzel / N.N.

764

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: **12 months**

participation according to Rili-BÄK:
 bi-annual
 (grace period: 30. May 2026)

CCR5-delta32

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - CFTR Common Mutations

Hentschel / Ahting

778

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 months / **24 months**

participation according to Rili-BÄK:
 yearly

CFTR gene ACMG classification Interpretation of results
 CFTR gene (NM_000492.4).

The 34 most common variants detected by standard screening methods will be tested. Participants must be able to identify at least the 34 most common variants. See here for a list of mandatory mutations: www.instand-ev.de/cftr-mandatory-mutations

Exception: Participants who only test the 31 variants of the child guidelines for CF. These participants are requested to communicate this in the comment field. Participants who only test for F508del cannot participate in this EQA scheme.

Participants who analyze the complete CFTR gene are also welcome to participate. These participants please communicate variants that are not among the 34 most common variants via the comment field. Participants are asked to comment on the possibility of CFTR modulator therapy.

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

Collagen receptor GP Ia/IIa (C 807 T)

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Congenital Adrenal Hyperplasia due to 21-Hydroxylase Deficiency (CYP21A2 Gene)

Christensen / Kappert

773

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 month / 24 months

Analysis of the CYP21A2 gene for small base variations (SNVs) and/or copy number variations (CNVs)

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Connexin 26 (GJB2 Gene, GJB6 Gene)

Christensen / Kappert

782

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 months / 24 months

participation according to Rili-BÄK:
yearly

Analysis of the GJB2 gene for small base variations (SNVs) and/or copy number variations (CNVs)

Analysis of the GJB6 gene for small base variations (SNVs) and/or copy number variations (CNVs)

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 months

participation according to Rili-BÄK:
 yearly

ATP 7 B: H 1069 Q

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - ctDNA Tumour Panel

Holdenrieder / N.N.

752

Samples : 2 samples of 3 ml (30 ng/ml cfDNA)
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual
 (grace period: 30. May 2026)

BRAF V600-variant(s) EGFR T790M-variant(s) ESR1-variant(s) KRAS Codon
 12-variant(s) KRAS Codon 13-variant(s) KRAS Codon 61-variant(s) NRAS Codon
 61-variant(s) PIK3CA-variant(s)

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - CYP2C9 CYP2C19 VKORC1 TPMT

Wenzel / N.N.

775

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months / 12 months

participation according to Rili-BÄK:
 bi-annual

CYP2C19*2 Allele: G 681 A CYP2C19*3 Allele: G 636 A CYP2C9*2 Allele: C 430 T
 CYP2C9*3 Allele: A 1075 C TPMT (nt. A 719 G) TPMT (nt. G 460 A)
 TPMT*2 Allele (nt. G 238 C) VKORC1: C 1173 T VKORC1: G -1639 A

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 2 samples of 40 µl Sample properties: DNA solution Shipping Information: Shipping at ambient temperature Certificate Validity: 12 months / 12 months				participation according to Rili-BÄK: bi-annual
CYP2D6 Gene Duplication complete Deletion	CYP2D6*3: A2637 del CYP2D6*6: T1795 del	CYP2D6*4: G 1934 A IL-28B C/T: rs 12979860	CYP2D6*5:	
		May (3)	Nov. (6)	
Registration deadline:		27.02.26	07.08.26	
Sample shipment:		28.04.26	06.10.26	
Closing Date:		29.05.26	06.11.26	

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months / 12 months

participation according to Rili-BÄK:
 bi-annual
 (grace period: 30. May 2026)

The following variants are tested in this EQA scheme: Haplotype B3 (c.1236G>A; c.1129-5923C>G); c.1679T>G; c.1905+1G>A; c.2846A>T and the assessment of the influence on enzyme function as well as the dose recommendation

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Duchenne Muscular Dystrophy (DMD) and Becker Muscular Dystrophy (BMD) (DMD Gene)

Christensen / Kappert

780

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 month / 24 months

participation according to Rili-BÄK:
 yearly

Analysis of the DMD gene for small base variations (SNVs) and/or copy number variations (CNVs)

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Factor V-Leiden-Gene (F5) - Whole Blood

Flörke / Kappert

730

Samples : 2 samples of 0.5 ml
 Sample properties: Whole blood
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Factor V-Leiden

	Apr. (2)	Oct. (5)
Registration deadline:	09.01.26	03.07.26
Sample shipment:	10.03.26	01.09.26
Closing Date:	10.04.26	02.10.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Factor V-Leiden

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

Fibrinogen Receptor HPA: 1a, 1b

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 months / 24 months

participation according to Rili-BÄK:
 yearly

FMR1: determination of CGG-repeats in 5' UTR

FMR1: determination of the variant category

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl Sample properties: DNA solution Shipping Information: Shipping at ambient temperature Certificate Validity: 12 months			participation according to Rili-BÄK: bi-annual
HFE: C 282 Y	HFE: H 63 D	HFE: S 65 C	
	May (3)	Nov. (6)	
Registration deadline:	27.02.26	07.08.26	
Sample shipment:	28.04.26	06.10.26	
Closing Date:	29.05.26	06.11.26	

Samples : 2 samples of 0.5 ml Sample properties: Whole blood Shipping Information: Shipping at ambient temperature Certificate Validity: 12 months			participation according to Rili-BÄK: bi-annual
HFE: C 282 Y	HFE: H 63 D	HFE: S 65 C	
	Apr. (2)	Oct. (5)	
Registration deadline:	09.01.26	03.07.26	
Sample shipment:	10.03.26	01.09.26	
Closing Date:	10.04.26	02.10.26	

Samples : 2 samples of 40 µl Sample properties: DNA solution Shipping Information: Shipping at ambient temperature Certificate Validity: 24 months / 24 months		participation according to Rili-BÄK: yearly
Determination of CAG-repeats		
Evaluation		
	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Lactase-Gene -13910T>C (MCM6 Gene, IVS13 C-T, rs4988235, HGVS: C.1917+326 C>T)

Christensen / Kappert

770

Samples : 2 samples of 40 µl
Sample properties: DNA solution
Shipping Information: Shipping at ambient temperature
Certificate Validity: 12 months

participation according to Rili-BÄK:
bi-annual

Laktase-Gene (LCT) (-13910 C>T; HGVS: c.1917+326C>T)

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - MLH1 MSH2 MSH6 PMS2 (Lynch)

Hentschel / Ahting

783

Samples : 2 samples of 40 µl
Sample properties: DNA solution
Shipping Information: Shipping at ambient temperature
Certificate Validity: 24 month / 24 months

participation according to Rili-BÄK:
yearly

MLH1, MSH2,MSH6,PMS2 gene analysis (SNVs und CNVs)

Classification of variants

Interpretation and recommendations

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

MTHFR (A 1298 C)

MTHFR (C 677 T)

May (3)

Nov. (6)

Registration deadline:

27.02.26

07.08.26

Sample shipment:

28.04.26

06.10.26

Closing Date:

29.05.26

06.11.26

Samples : 2 samples of 0.5 ml
 Sample properties: Whole blood
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

MTHFR (A 1298 C)

MTHFR (C 677 T)

Apr. (2)

Oct. (5)

Registration deadline:

09.01.26

03.07.26

Sample shipment:

10.03.26

01.09.26

Closing Date:

10.04.26

02.10.26

Samples : 2 samples of 1.5 ml
 Sample properties: Liquid plasma (Plasma)
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

Determination of the fetal RhD status

Shipping to Germany, Austria and Swiss only.

May (3)

Oct. (6)

Registration deadline:

27.02.26

07.08.26

Sample shipment:

28.04.26

06.10.26

Closing Date:

07.05.26

16.10.26

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

NOD2 mutation(s) NOD2 Nomenclature cDNA

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - PAI-1-Gene (SERPINE1) (4G/5G)

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

Plasminogenactivator-Inhibitor (PAI-1): 4G, 5G

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - PAI-1-Gene (SERPINE1) (4G/5G) - whole blood

Samples : 2 samples of 0.5 ml
 Sample properties: Whole blood
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

Plasminogenactivator-Inhibitor (PAI-1): 4G, 5G

	Apr. (2)	Oct. (5)
Registration deadline:	09.01.26	03.07.26
Sample shipment:	10.03.26	01.09.26
Closing Date:	10.04.26	02.10.26

Molecular Genetics - Prader-Willi (PWS) und Angelman (AS) Syndromes (Chromosome region 15q11-13, SNRPN)

Christensen / Kappert

786

Samples : 2 samples of 40 µl
Sample properties: DNA solution
Shipping Information: Shipping at ambient temperature
Certificate Validity: 24 month / **24 months**

participation according to Rili-BÄK:
yearly

Methylation analysis

Deletion analysis

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Protein C (PROC Gene)

Christensen / Kappert

790

Samples : 2 Samples of 50 µl
Sample properties: DNA solution
Shipping Information: Shipping at ambient temperature
Certificate Validity: 12 months

Analysis of the PROC gene for small base variations (SNVs) and/or copy number variations (CNVs)

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Molecular Genetics - Protein S (PROS1 Gene)

Christensen / Kappert

791

Samples : 2 Samples of 50 µl
Sample properties: DNA solution
Shipping Information: Shipping at ambient temperature
Certificate Validity: 12 months

Analysis of the PROS1 gene for small base variations (SNVs) and/or copy number variations (CNVs)

Interpretation of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Prothrombin G20210A

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 2 samples of 0.5 ml
 Sample properties: Whole blood
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

participation according to Rili-BÄK:
 bi-annual

Prothrombin G20210A

	Apr. (2)	Oct. (5)
Registration deadline:	09.01.26	03.07.26
Sample shipment:	10.03.26	01.09.26
Closing Date:	10.04.26	02.10.26

Samples : 2 samples of 40 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 24 month / 24 months

participation according to Rili-BÄK:
 yearly

Determination of the copy numbers of the SMN1 and SMN2 genes

Interpretations of results

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26

Samples : 3 samples of 20 µl
 Sample properties: DNA solution
 Shipping Information: Shipping at ambient temperature
 Certificate Validity: 12 months

**participation according to Rili-BÄK:
 bi-annual**

UDP-glucuronyl transferase (TA)6, (TA)7

	May (3)	Nov. (6)
Registration deadline:	27.02.26	07.08.26
Sample shipment:	28.04.26	06.10.26
Closing Date:	29.05.26	06.11.26