

Liste der Untersuchungsverfahren im flexiblen Geltungsbereich

Herausgeber: Institut für Humangenetik
 Geltungsbereich: Institut für Humangenetik, MVZ-Humangenetik
 Berufsgruppe: Ärztlicher Dienst, Wissenschaftliche Mitarbeitende
 Revision: 006/16.05.2024
 Dok.-ID: 79182
 Freigabe abgeschlossen am: 16.05.2024
 Freigabe abgeschlossen durch: Alter, Svenja

Untersuchungsgebiet: Humangenetik										
Analyt (Meßgröße)	Untersuchungsmaterial (Matrix)	Untersuchungstechnik	Anweisung/Version	Gerät	CE-Verfahren	Inhouse-Verfahren	akkreditiert	Flexibler Geltungsbereich	letzte Änderung	Panel-ID
Whole Exome Sequencing (~ 20.000 humane, protein-kodierende Gene)	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience), Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	SOP_UV_Whole Exome Sequencing/001	Illumina NextSeq1000	nein	ja	ja	ja	20.02.2023	-
Häufige Ichthyosen	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_01
Autosomal rezessive Ichthyosen	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	SOP_UV_Autosomal rezessive Ichthyosen/001	Illumina MiSeq, Illumina NextSeq1000	nein	ja	ja	ja	01.03.2023	SKIN_02
Autosomal dominante Ichthyose	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_03
Keratinopathische Ichthyosen	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	SOP_UV_Keratinopathische Ichthyosen/001	Illumina MiSeq, Illumina NextSeq1000	nein	ja	ja	ja	23.02.2023	SKIN_04
Syndromale Ichthyosen	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_05
Hyperkeratosen und Peeling-Skin-Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_06
Erythrokeratoderma variabilis	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	07.12.2022	SKIN_26
Diffuse PPK	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_07
Striäre PPK	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_08
Punktierte PPK	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_09
Pachyonychia congenital	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_10
Dysplasien mit assoziierter PPK	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_11
Sonstige Erkrankungen mit PPK	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_12
Porokeratose	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_13
Loricrin-Keratoderma	EDTA-Blut, DNA; DNA	PCR und Sanger-Sequenzierung	Sanger-Sequenzierung/001	ABI Kapillarsequenzierer 3130, ABI Kapillarsequenzierer 3500	nein	ja	nein	-	08.02.2023	SKIN_27
Epidermolysis bullosa simplex	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_14
Epidermolysis bullosa dystrophica	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_15
Epidermolysis bullosa junctionalis	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_16
Kindler-Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_17
Keratinozytärer Nävus	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_18
Epidermolytischer Nävus	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_19
Nävus sebaceus	EDTA-Blut, DNA; DNA	Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina MiSeq, Illumina NextSeq1000	nein	ja	nein	-	30.05.2022	SKIN_20

epileptische Enzephalopathien	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_02
Dravet-Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_03
Generalisierte und fokale Epilepsien	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_04
nächtliche Frontallappenepilepsie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_05
syndromale Epilepsie (ohne schwere Enzephalopathie)	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_06
Generalisierte Epilepsie mit Fieberkrämpfen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_07
Epilepsie und X-gekoppelte mentale Retardierung	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_08
generalisierte myoklonische Epilepsien	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_09
Epilepsien mit höherer Therapierelevanz	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_10
Hyperekplexie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_11
Gehirnfehlbildungen (Lissenzephalien, Tubulinopathien, pontozebelläre Hypoplasie)	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	EH_12
Charcot-Marie-Tooth-Erkrankung Typ 1 und Typ 2 bzw. HNPP; 1. Stufe	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	NP_01
motorisch-sensible Neuropathie, demyelinisierend (CMT1)	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	27.06.2022	NP_02
motorisch-sensible Neuropathie, axonal (CMT2)	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	29.06.2022	NP_03
motorische Neuropathie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	NP_04
sensorische/autonome Neuropathie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	29.06.2022	NP_05
Parkinson	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	07.07.2022	ND_01
Demenz (inkl. Alzheimer, frontotemporale Demenz)	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	ND_02
Leukenzephalopathie und Leukodystrophie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	ND_03
Amyotrophe Lateralsklerose	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	ND_04
Neuronale Ceroid-Lipofuszinose	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	ND_05
Ataxie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	ND_06
spastische Spinalparalyse	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	ND_07
Dystonie und paroxysmale Bewegungsstörungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	ND_08
Autismus	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	30.06.2022	INT_01
Leukodystrophien	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	INT_02
Strukturelle Fehlbildungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	29.06.2022	INT_03
Lysosomale Erkrankungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	INT_04
Faziale Dismorphie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MiSeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	INT_05

Mikrozephalie	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	30.06.2022	INT_06
Sonstige Gene Intelligenzminderung	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	29.06.2022	INT_07
Zystische Nierenerkrankungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	REN_01
Nierenfehlentwicklung	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	REN_02
Glomerulopathien	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	REN_03
Tubuläre Erkrankungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	REN_04
Endokrine Nierenerkrankungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	REN_05
Renale Tumorsyndrome	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	18.05.2022	REN_06
Nephrotisches Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	10.05.2023	REN_07
Kleinwuchs	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	BONE_01
Erkrankungen mit Extremitätenfehlbildungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	BONE_02
Erkrankungen mit reduzierter Knochendichte	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	BONE_03
Erkrankungen mit erhöhter Knochendichte	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	BONE_04
Kraniosynostosen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	BONE_05
Skeletterkrankungen mit gestörter Mineralisation	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	BONE_06
Weitere Skelettdysplasien und ausgewählte Syndrome mit Skelettbeteiligung	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	19.05.2022	BONE_07
Störungen des Cholesterinstoffwechsels	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	LIPID_01
Störungen des Triglyzeridstoffwechsels	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	LIPID_02
sonstige Lipidstoffwechselfstörungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	LIPID_03
Periodisches Fieber / Klassische autoinflammatorische Syndrome	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	AUTOINFLAM_01
Panel für weitere autoinflammatorische Erkrankungen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	AUTOINFLAM_02
Chilblain Lupus erythematodes	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	AUTOINFLAM_03
Panel für weitere Interferonopathien	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	AUTOINFLAM_04
Immundefizienzen	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	AUTOINFLAM_05
Hämophagozytische Lymphohistiozytose / lymphoproliferatives Syndrom	EDTA-Blut, DNA; DNA	Sequence capture (Twist Human Core Exome Enrichment, Fa. Twist Bioscience) Sequencing-by-Synthesis (Illumina NextSeq1000), inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	NGS_Panel_MISeq002 NGS_Panel_NextSeq003	Illumina NextSeq1000	nein	ja	nein	-	24.05.2022	AUTOINFLAM_06
Zystische Fibrose (CFTR)	EDTA-Blut, DNA; DNA	PCR und Sanger-Sequenzierung, MiPA, Sequence capture (Custom Panel Fa. Agilent, Custom Panel, Fa. Twist Bioscience), Sequencing-by-Synthesis, inhouse-pipeline, commercial pipeline (JSI medical systems GmbH v5.2.0)	SOP_UV_Zystische Fibrose/001	Illumina MiSeq, Illumina NextSeq1000, ABI Kapillarsequenzierer 3130, ABI Kapillarsequenzierer 3500	nein	ja	ja	ja	01.03.2023	
Huntington-Erkrankung (HTT)	EDTA-Blut, DNA; DNA	Short Tandem Repeat typing	SOP_UV_Huntington-Erkrankung/002	ABI Kapillarsequenzierer 3130, ABI Kapillarsequenzierer 3500	nein	ja	ja	ja	06.03.2023	
Fuchs-Endotheldystrophie (TCF4 Repeatanalyse)	EDTA-Blut, DNA; DNA	Short Tandem Repeat typing	Fragmentlängenanalyse002	ABI Kapillarsequenzierer 3130, ABI Kapillarsequenzierer 3500	nein	ja	nein	-	16.11.2023	
Aderhautmelanom	EDTA-Blut, DNA, DNA aus Tumorgewebe; DNA	PCR und Sanger-Sequenzierung an DNA aus Tumorgewebe (GNAQ und GNA11, jeweils Exon 4 und 5) sowie Short Tandem Repeat typing an DNA aus EDTA-Blut sowie an DNA aus Tumorgewebe	Fragmentlängenanalyse002	ABI Kapillarsequenzierer 3130, ABI Kapillarsequenzierer 3500	nein	ja	nein	-	16.11.2023	