



Pharmacogenetic Tests

Prices of the tests are in Euro, but can be converted to your local currency with the [currency converter](#).

More information on our Pharmacogenetic Tests is available from our website www.pharmaco-GENDIA.net.

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Pharmacogenetic Tests offered:

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Oncology

Pharmacogenetic tests used in oncology can be divided in 2 groups:

1. Tests determining the toxicity of chemotherapeutics such as 5-Fluorouracil, Irinotecan and Thiopurine. Such toxicity is caused by mutations in the genes encoding Dihydropyrimidine Dehydrogenase, UDP-Glycucuronosyl transferase or Thiopurine S-Methyltransferase, respectively.
2. Tests determining the response to the treatment with specific tyrosine kinase inhibitors (TKIs). Such response is determined by mutations in specific tyrosine kinase genes. Only patients with a mutation in the specific tyrosine kinase will respond to treatment with the specific TKI. Also the development of resistance against TKIs is genetically determined by mutations in the tyrosine kinase genes.

The most prominent examples of pharmacogenetic tests used in oncology are given below.

These tests require samples specified in the column: Tissue

Test	Disease	Gene	Tissue	Test Number	Price in Euro
5-FLUORO URACIL TOXICITY	VARIOUS	ALLELE 2A (IVS14+1G-A) IN DPD (DIHYDROPYRIMIDINE DEHYDROGENASE)	DNA	1	200
		ALLELES *3,*4, *5A, *7, *8, *9, *10, *12, *13, M166V, R886H, D949V IN DPD (DIHYDROPYRIMIDINE DEHYDROGENASE)	DNA	2	800
IRINOTECAN TOXICITY	VARIOUS	TA INSERTION IN PROMOTOR OF UGT1A1 (UDP-GLYCUCURONOSYL TRANSFERASE)	DNA	3	195
THIOPURINE TOXICITY	VARIOUS	ALLELES 1, 2, 3A, 3C IN TMPT (THIOPURINE S-METHYLTRANSFERASE)	DNA	4	130
HERCEPTIN RESPONSIVENESS	BREAST CANCER	HER2 / NEU OVEREXPRESSION	PARAFFINISED BREAST TUMOUR TISSUE	5	380
GLEEVEC / IMATINIB RESPONSIVENESS	CHRONIC MYELOGENOUS LEUKEMIA (CML) AND ACUTE LEUKEMIA	EXONS 4-10 MUTATIONS IN ABL (INCLUDING T315I)	BLOOD OR BONE MARROW IN PAX RNA TUBES	6	440
GLEEVEC / IMATINIB RESPONSIVENESS	CHRONIC MYELOGENOUS LEUKEMIA (CML) AND ACUTE LEUKEMIA	FUSION OF ABL TO BCR	BLOOD OR BONE MARROW IN PAX RNA TUBES	7	200
GLEEVEC / IMATINIB RESPONSIVENESS	ACUTE LEUKEMIA	FUSION OF PDGFRB TO TEL/ETV6	BLOOD OR BONE MARROW IN PAX RNA TUBES	8	200
GLEEVEC / IMATINIB RESPONSIVENESS	HYPEREOSINOPHILIC SYNDROME	FUSION OF PDGFRB TO TEL/ETV6	BLOOD OR BONE MARROW IN PAX RNA TUBES	8	200
GLEEVEC / IMATINIB RESPONSIVENESS	HYPEREOSINOPHILIC SYNDROME	del(4)(q12q12) WITH FIP1L1-PDGFR A FUSION	BLOOD OR BONE MARROW IN PAX RNA TUBES	9	300
GLEEVEC / IMATINIB RESPONSIVENESS	CHRONIC EOSINOPHILIC LEUKEMIA	del(4)(q12q12) WITH FIP1L1-PDGFR A FUSION	BLOOD OR BONE MARROW IN PAX RNA TUBES	9	300
GLEEVEC / IMATINIB RESPONSIVENESS	ACUTE MYELOID LEUKEMIA	MUTATIONS IN EXONS 8, 11 AND 17 IN KIT	WHOLE BLOOD, BONE MARROW ASPIRATE OR PARAFFIN-EMBEDDED BIOPSY	10	500

GLEEVEC / IMATINIB RESPONSIVENESS	MASTOCYTOSIS	MUTATIONS IN EXON 17 IN KIT	PARAFFINISED TUMOUR TISSUE	11	300
GLEEVEC / IMATINIB RESPONSIVENESS	MAST CELL LEUKEMIA	MUTATIONS IN EXON 17 IN KIT	WHOLE BLOOD, BONE MARROW ASPIRATE OR PARAFFIN-EMBEDDED BIOPSY	11	300
GLEEVEC / IMATINIB RESPONSIVENESS	GASTROINTESTINAL STROMAL TUMOR, GIST	MUTATIONS IN EXONS 9, 11, 13 AND 17 IN KIT	PARAFFINISED TUMOUR TISSUE	12	600
GLEEVEC / IMATINIB RESPONSIVENESS	GASTROINTESTINAL STROMAL TUMOR, GIST	MUTATIONS IN EXONS 12 AND 18 IN PDGFRA	PARAFFINISED TUMOUR TISSUE	13	400
IRESSA / GEFITINIB RESPONSIVENESS	NON SMALL CELL LUNG CANCER (NSCLC)	EXON 18-21 MUTATIONS IN EGFR (EPIDERMAL GROWTH FACTOR RECEPTOR)	FRESH TISSUE (ETHANOL-FIXED TISSUE)	14	630
VARIOUS FLT3 INHIBITORS	ACUTE MYELOID LEUKEMIA	ACTIVATING MUTATION (INTERNAL TANDEM DUPLICATION) IN FLT3 (RECEPTOR TYROSINE KINASE)	BLOOD OR BONE MARROW	15	300
VARIOUS FLT3 INHIBITORS	ACUTE MYELOID LEUKEMIA	ACTIVATING MUTATIONS IN EXON 14 IN FLT3 (RECEPTOR TYROSINE KINASE)	BLOOD OR BONE MARROW	16	200
BETA2-AGONISTS RESPONSE	VARIOUS	R16G AND Q27E MUTATIONS IN ADRB2	DNA	18	310
ABACAVIR TOXICITY	VARIOUS	HLA-B*5701	DNA	17	220

Other Disciplines

More than 30 genes are involved in the metabolism of drugs. Mutations in these genes determine variation in the enzyme activity leading to poor, intermediate, fast or ultrafast breakdown and excretion of many drugs.

Poor metabolisers are at risk for adverse drug reactions, whereas the efficacy of the medication in ultrafast metabolisers is reduced.

The most prominent examples of such pharmacogenetic tests used are given in the table below.

Gene	Alleles*	Effect	Test Number	Price in Euro
CYP2D6	*3, *4, *5, *6, *7, *8, *9, *14, *19	Poor metaboliser	19	Upon Request
	*XN	Ultrafast metaboliser	20	Upon Request
	*3, *4, *5, *6, *7, *8, *9, *14, *19, *XN	Poor and ultrafast metaboliser	21	Upon Request
CYP2D6/CYP2C19	AMPLICHIP CYP450 WITH 34 ALLELES OF CYP2D6/CYP2C19: CYP2D6 alleles: *2, *3, *4, *5, *6, *7, *8, *9, *10, *11, *14A, *14B, *15, *17, *19, *20, *25, *26, *29, *30, *31, *35, *36, *40, *41, 1XN, 2XN, 4XN, 10XN, 17XN, 35XN, 41XN CYP2C19 alleles: *2, *3	Poor and ultrafast metaboliser	22	Upon Request
CYP2C19	*2, *2B, *3, *4, *5, *6, *7, *8, *9, *10, *11	Poor metaboliser	23	Upon Request
CYP2C9	*2, *3, *4, *5, *6, *11	Poor metaboliser	24	Upon Request

* For detailed information on mutations and additional tests see table below.

A large amount of variations have been described in the genes encoding phase I and phase II enzymes. A complete table of all variations offered through GENDIA is listed below.

Gene	Allele	Mutation	Effect on Protein	Effect on Enzyme Activity*	Test Number	Price in Euro	
Cytochrome P450 (CYP)							
CYP1A2	*1C	g.-3858G>A		decreased activity	25	Upon Request	
	*1E	g.-740T>G			26	Upon Request	
	*1F	g.-164C>A		higher inducibility	27	Upon Request	
	*1J	g.-740T>G g.-164C>A			28	Upon Request	
	*1K	g.-740T>G g.-730C>T g.-164C>A			decreased activity	29	Upon Request
	*1E, *1F, *1J, *1K					30	Upon Request
	*2	g.63C>G	F21L			31	Upon Request
	*3	g.2116G>A	D348N			32	Upon Request
	*4	g.2499A>G	I348N			33	Upon Request
	*5	g.3497G>A	C406Y			34	Upon Request
	*6	g.5090C>T	R431W			35	Upon Request

	*7	g.3534G>A	splicing defect	decreased activity	36	Upon Request	
CYP2A6	*1H	g.-745A>G			37	Upon Request	
	*2	c.479T>A	L160H	no activity	38	Upon Request	
	*4	deletion	no protein	no activity	39	Upon Request	
	*5	c.1436G>T	G479V	no activity	40	Upon Request	
	*6	c.383G>A	R128Q	decreased activity	41	Upon Request	
	*7	c.1412T>G	I471T	decreased activity	42	Upon Request	
	*9	c.-48T>G	decreased protein	decreased activity	43	Upon Request	
	*10	c.1412G>T c.1454G>T	I471T, R485L	decreased activity	44	Upon Request	
	*11	c.670T>C	S224P	decreased activity	45	Upon Request	
	*12	partial deletion	altered protein	decreased activity	46	Upon Request	
	*17	c.1093G>A	V365M	decreased activity	47	Upon Request	
	*1x2	gene duplication	increased protein	increased activity	48	Upon Request	
	CYP2B6	*2	c.64C>T	R22C		49	Upon Request
		*3	c.777C>A	S259R		50	Upon Request
*4		c.785A>G	K262R		51	Upon Request	
*5		c.1459C>T	R487C		52	Upon Request	
*6		c.516G>T c.785A>G	Q172H, K262R		53	Upon Request	
*7		c.516G>T c.785A>G c.1459C>T	Q172H, K262R, R487C		54	Upon Request	
*8		c.415A>G	K139E	decreased activity	55	Upon Request	
*9		c.516G>T	Q172H		56	Upon Request	
CYP2C8		*2	c.805A>T	I269F	increased activity	57	Upon Request
	*3	c.416G>A c.1196A>G	R139K, K399R	decreased activity	58	Upon Request	
	*4	c.792C>G	I264M		59	Upon Request	
	*5	c.475delA	T159fs177X	no activity	60	Upon Request	
	*7	c.556C>T	R186X	no activity	61	Upon Request	
	*8	c.556C>G	R186G	decreased activity	62	Upon Request	
	CYP2C9	*2	c.430C>T	R144C	decreased activity	63	Upon Request
		*3	c.1075A>C	I359L	decreased activity	64	Upon Request
*4		c.1076T>C	I359T	decreased activity	65	Upon Request	
*5		c.1080C>G	D360E	decreased activity	66	Upon Request	
*6		c.818delA	frameshift	no activity	67	Upon Request	
*7		c.55C>A	L19I		68	Upon Request	
*8		c.449G>A	R150H	increased activity	69	Upon Request	
*9		c.752A>G	H251R		70	Upon Request	
*10		c.815A>G	E272G		71	Upon Request	
*11		c.1003C>T	R335W	decreased activity	72	Upon Request	
*12		c.1465C>T	P489S	decreased activity	73	Upon Request	
*16		c.485C>A	T299A	decreased activity	74	Upon Request	
*18		c.1075A>C	I359L	decreased activity	75	Upon Request	
		*2, *3, *4, *5, *6, *11			poor metaboliser	24	Upon Request
CYP2C19	*2	c.681G>A	splicing defect	no activity	76	Upon Request	
	*3	c.636G>A	W212X	no activity	77	Upon Request	
	*4	c.1A>G	start codon mutation	no activity	78	Upon Request	
	*5	c.1297G>A	R433W	no activity	79	Upon Request	

	*6	c.395G>A	R132Q	no activity	80	Upon Request
	*7	1VS5+2T>A	splicing defect	no activity	81	Upon Request
	*8	c.358T>C	W120R	no activity	82	Upon Request
	*9	c.431G>A	R144H	decreased activity	83	Upon Request
	*10	680C>T	P227L	decreased activity	84	Upon Request
	*11	c.449G>A	R150H		85	Upon Request
	*2, *2B, *3, *4, *5, *6, *7, *8, *9, *10, *11			poor metaboliser	23	Upon Request
CYP2D6	*3	g.2549delA	frameshift	no activity	86	Upon Request
	*3B	g.1749A>G g.2549delA	N166D, frameshift	no activity	87	Upon Request
	*4	g.1846G>A	splicing defect	no activity	88	Upon Request
	*5	gene deletion	no protein	no activity	89	Upon Request
	*6	g.1701delT	frameshift	no activity	90	Upon Request
	*7	g.2935A>C	H374P	no activity	91	Upon Request
	*8	g.1758G>T	Stop codon	no activity	92	Upon Request
	*9	g.2613-2615delAGA	K281del	decreased activity	93	Upon Request
	*14	g.1758G>A	G169R	no activity	94	Upon Request
	*17	g.1023C>T g.2850C>T	T107I, R296C	decreased activity	95	Upon Request
	*20	g.1973insG	frameshift	no activity	96	Upon Request
	*21	g.2573insC	frameshift	no activity	97	Upon Request
	*24	g.2853A>C	I297L		98	Upon Request
	*38	g.2587-2590delGACT	frameshift	no activity	99	Upon Request
	*44	g.2950G>C	splicing defect	no activity	100	Upon Request
	*XN	gene amplification	increased protein	increased activity	20	Upon Request
		*3, *4, *5, *6, *7, *8, *9, *14, *19			poor metaboliser	19
	*3, *4, *5, *6, *7, *8, *9, *14, *19, *XN			poor and ultrafast metaboliser	21	Upon Request
CYP2D6 / CYP2C19 AmpliChip (Roche Diagnostics)	*2, *3, *4, *5, *6, *7, *8, *9, *10, *11, *14A, *14B, *15, *17, *19, *20, *25, *26, *29, *30, *31, *35, *36, *40, *41, 1XN, 2XN, 4XN, 10XN, 17XN, 35XN, 41XN + CYP2C19				22	Upon Request
CYP2E1	*1C	6 minisat			101	Upon Request
	*1D	8 minisat		increased activity	102	Upon Request
	*2	g.1132G>A	R76H	decreased activity	103	Upon Request
	*5	g.-1293G>C g.-1053C>T			104	Upon Request
	*6	g.7632T>A			105	Upon Request
	*1C, *1D				106	Upon Request
CYP2J2	*2	c.427A>G	T143A	decreased activity	107	Upon Request
	*3	c.472C>T	R158C	decreased activity	108	Upon Request
	*4	c.575T>A	I192N	decreased activity	109	Upon Request
	*5	c.1024G>A	D342N	decreased activity	110	Upon Request
	*6	c.1210A>T	N404Y	decreased activity	111	Upon Request
	*7	g.-76G>T	decreased protein	decreased activity	112	Upon Request
CYP3A4	*1B	g.-392A>G			113	Upon Request
	*2	g.15713T>C	S222P	decreased activity	114	Upon Request
	*3	g.23172T>C	M445T		115	Upon Request

	*4	g.13871A>G	I118V		116	Upon Request
	*5	g.15702C>G	P218R		117	Upon Request
	*6	g.17662-17663insA	frameshift		118	Upon Request
	*7	g.6004G>A	G56D		119	Upon Request
	*8	g.13908G>A	R130Q		120	Upon Request
	*9	g.14292G>A	V170I		121	Upon Request
	*10	g.14304G>C	D174H		122	Upon Request
	*11	g.21867C>T	T363M		123	Upon Request
	*12	g.21896C>T	L373F		124	Upon Request
	*13	g.22026C>T	R416L		125	Upon Request
	*14	g.44T>C	L15P		126	Upon Request
	*15	g.14269G>A	R162Q		127	Upon Request
	*16	g.15603C>G	T185S		128	Upon Request
	*17	g.15615T>C	F189S	decreased activity	129	Upon Request
	*18	g.20070T>C	L293P	increased activity	130	Upon Request
	*19	g.23237C>T	P467S		131	Upon Request
CYP3A5	*2	g.27289C>A	T398N		132	Upon Request
	*3C	g.6986G>A	splicing defect	no activity	133	Upon Request
	*4	g.14665A>G	Q200R		134	Upon Request
	*5	g.12952T>C	splicing defect		135	Upon Request
	*6	g.14690G>A	splicing defect	no activity	136	Upon Request
	*7	g.27131-27132insT	frameshift		137	Upon Request
	*8	g.3699C>T	R28C	decreased activity	138	Upon Request
	*9	g.19386G>A	A337T	decreased activity	139	Upon Request
	*10	g.29753T>C	splicing defect	decreased activity	140	Upon Request
CYP3A7	*1C	c.-291G>T -284T>A -282T>C -281A>T -270T>G -262T>A -232A>C		increased activity	141	Upon Request
	*2	c.1226C>G	T409R	increased activity	142	Upon Request
CYP4B1	*2	c.881-882delAT	frameshift		143	Upon Request
Epoxidhydroxylases (EPHX)						
EPHX1	n.a.	c.128G>C	R43T		144	Upon Request
	*3	c.337T>C	Y113H		145	Upon Request
	*4	c.416A>G	H139R		146	Upon Request
EPHX2	n.a.	c.229A>G	K55R	increased activity	147	Upon Request
	n.a.	c.307C>T	R103C		148	Upon Request
	n.a.	c.461G>A	C154	increased activity	149	Upon Request
	n.a.	c.860G>A	R287Q	decreased activity	150	Upon Request
	n.a.	c.1208-1209insTCG	403-404insR	decreased activity	151	Upon Request
Glutathione S-transferases (GST)						
GSTM1	*0	gene deletion	no protein		152	Upon Request
GSTT1	*A	wild type			153	Upon Request
	*B	c.301A>C	T104P		154	Upon Request
	*0	gene deletion	no protein		155	Upon Request
GSTP1	*A	wild type			156	Upon Request
	*B	c.313A>G	I105V		157	Upon Request

	*C	c.313A>G c.341C>T	I105V, A114V		158	Upon Request
Sulfonyl transferases (SULT)						
SULT1A1	*2	638 G>A	R213H	decreased activity	159	Upon Request
	*3	667 G>A	M223V		160	Upon Request
	*2, *3				161	Upon Request
SULT1A2	*2	20 T>C	I7T		162	Upon Request
	*3	56 T>C	P19L		163	Upon Request
	*2, *3				164	Upon Request
N-Acetyltransferase type 2 (NAT2)						
NAT2	*4	wild type			165	Upon Request
	*5	c.341T>C	I114T	decreased activity	166	Upon Request
	*6	c.590G>T	R197Q	decreased activity	167	Upon Request
	*7	c.857G>A	G286E	decreased activity	168	Upon Request
	*10	c.499G>A	E167K		169	Upon Request
	*11B	c.481C>T;859del	frameshift		170	Upon Request
	*12	c.803A>G	K268R		171	Upon Request
	*13	c.282C>T	none		172	Upon Request
	*14	c.191G>A	R64Q	decreased activity	173	Upon Request
	*17	c.434A>C	Q145P	decreased activity	174	Upon Request
	*18	c.845A>C	K282T		175	Upon Request
	*19	c.190C>T	R64W		176	Upon Request
		*4, *5, *6, *7, *10, *11B, *12, *13, *14, *17, *18, *19				177
Thiopurine methyltransferases (TPMT)						
TPMT	*2	c.288G>C	A80P	decreased activity	178	Upon Request
	*3A	c.460G>A c.719A>G	A154T, Y240C	decreased activity	179	Upon Request
	*3B	c.719A>G	Y240C	decreased activity	180	Upon Request
	*3C	c.719A>G	A154T	decreased activity	181	Upon Request
Uridine diphosphate-glucuronyltransferases (UGT)						
UGT1A1	*1	promoter repeat [TA] 6	wild type		182	Upon Request
	*6	226A>G	G71R	decreased activity	183	Upon Request
	*28	promoter repeat [TA] 7		decreased activity	184	Upon Request
	*36	promoter repeat [TA] 5		increased activity	185	Upon Request
	*37	promoter repeat [TA] 8		decreased activity	186	Upon Request
		*28, **36, *37				187
UGT1A6	*1		wild type		188	Upon Request
	*2	637A>G 648A>C	T181A, R184S	decreased activity	189	Upon Request
UGT1A7	*1		wild type		190	Upon Request
	*2	387T>G 391C>A 392G>A	N129K, R131K	decreased activity	191	Upon Request
	*3	387T>G 391C>A 392G>A 622T>C	N129K, R131K, W208R	decreased activity	192	Upon Request
	*4	622T>C	W208R	decreased activity	193	Upon Request
		*1, *2, *3, *4				194
UGT2B4	*2	1411T>A	D458E	no activity	195	Upon Request
UGT2B7	*2	816C>T	H268Y	decreased activity	196	Upon Request
UGT2B15	*2	276T>G	D85Y	increased activity on androgens	197	Upon Request
Multidrug resistance 1 (MDR1, ABCB1)						

	n.a.	1236C>T	decreased protein	decreased activity	198	Upon Request
	n.a.	2677G>T or 2677G>A	A893S or A893T	decreased activity	199	Upon Request
	n.a.	3435C>T	decreased protein	decreased activity	200	Upon Request
5-@Hydroxytryptamine transporter (5-HTT, SLC6A4)						
	L, S	44 bp insertion/deletion in the promotor region	increased protein for L (insertion) variant	increased activity for L variant	201	Upon Request

*: only known effects on protein effect are given
n.a.: not applicable

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