



**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](http://www.pharmaco-GENDIA.net).

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**Pharmacogenetic Tests offered:**

- [Oncology](#)
- [Other Disciplines](#)

**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
<b>5-FLUORO URACIL TOXICITY</b>	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 IN DPD (DIHYDROPYRIMIDINE DEHYDROGENASE)	DNA	2	700
<b>IRINOTECAN TOXICITY</b>	VARIOUS	TA INSERTION IN PROMOTOR OF UGT1A1 (UDP-GLYCUCURONOSYL TRANSFERASE)	DNA	3	195
<b>THIOPURINE TOXICITY</b>	VARIOUS	ALLELES 1, 2, 3A, 3C IN TMPT (THIOPURINE S-METHYLTRANSFERASE)	DNA	4	130
<b>HERCEPTIN RESPONSIVENESS</b>	BREAST CANCER	HER2 / NEU OVEREXPRESSION	PARAFFINISED BREAST TUMOUR TISSUE	5	380
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	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
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	CHRONIC MYELOGENOUS LEUKEMIA (CML) AND ACUTE LEUKEMIA	FUSION OF ABL TO BCR	BLOOD OR BONE MARROW IN PAX RNA TUBES	7	200
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	ACUTE LEUKEMIA	FUSION OF PDGFRB TO TEL/ETV6	BLOOD OR BONE MARROW IN PAX RNA TUBES	8	200
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	HYPEREOSINOPHILIC SYNDROME	FUSION OF PDGFRB TO TEL/ETV6	BLOOD OR BONE MARROW IN PAX RNA TUBES	8	200
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	HYPEREOSINOPHILIC SYNDROME	del(4)(q12q12) WITH FIP1L1-PDGFR A FUSION	BLOOD OR BONE MARROW IN PAX RNA TUBES	9	300
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	CHRONIC EOSINOPHILIC LEUKEMIA	del(4)(q12q12) WITH FIP1L1-PDGFR A FUSION	BLOOD OR BONE MARROW IN PAX RNA TUBES	9	300
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	ACUTE MYELOID LEUKEMIA	MUTATIONS IN EXONS 8, 11 AND 17 IN KIT	WHOLE BLOOD, BONE MARROW ASPIRATE OR PARAFFIN-EMBEDDED BIOPSY	10	500

<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	MASTOCYTOSIS	MUTATIONS IN EXON 17 IN KIT	PARAFFINISED TUMOUR TISSUE	11	300
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	MAST CELL LEUKEMIA	MUTATIONS IN EXON 17 IN KIT	WHOLE BLOOD, BONE MARROW ASPIRATE OR PARAFFIN-EMBEDDED BIOPSY	11	300
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	GASTROINTESTINAL STROMAL TUMOR, GIST	MUTATIONS IN EXONS 9, 11, 13 AND 17 IN KIT	PARAFFINISED TUMOUR TISSUE	12	600
<b>GLEEVEC / IMATINIB RESPONSIVENESS</b>	GASTROINTESTINAL STROMAL TUMOR, GIST	MUTATIONS IN EXONS 12 AND 18 IN PDGFRA	PARAFFINISED TUMOUR TISSUE	13	400
<b>IRESSA / GEFITINIB RESPONSIVENESS</b>	NON SMALL CELL LUNG CANCER (NSCLC)	EXON 18-21 MUTATIONS IN EGFR (EPIDERMAL GROWTH FACTOR RECEPTOR)	FRESH TISSUE (ETHANOL-FIXED TISSUE)	14	720
<b>VARIOUS FLT3 INHIBITORS</b>	ACUTE MYELOID LEUKEMIA	ACTIVATING MUTATION (INTERNAL TANDEM DUPLICATION) IN FLT3 (RECEPTOR TYROSINE KINASE)	BLOOD OR BONE MARROW	15	300
<b>VARIOUS FLT3 INHIBITORS</b>	ACUTE MYELOID LEUKEMIA	ACTIVATING MUTATIONS IN EXON 14 IN FLT3 (RECEPTOR TYROSINE KINASE)	BLOOD OR BONE MARROW	16	200
<b>BETA2-AGONISTS RESPONSE</b>	VARIOUS	R16G AND Q27E MUTATIONS IN ADRB2	DNA	18	310
<b>ABACAVIR TOXICITY</b>	VARIOUS	HLA-B*5701	DNA	19	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
<b>CYP2D6</b>	*3, *4, *5, *6, *7, *8, *9, *14, *19	Poor metaboliser	19	230
	*XN	Ultrafast metaboliser	20	80
	*3, *4, *5, *6, *7, *8, *9, *14, *19, *XN	Poor and ultrafast metaboliser	21	270
<b>CYP2D6/CYP2C19</b>	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	900
<b>CYP2C19</b>	*2, *2B, *3, *4, *5, *6, *7, *8, *9, *10, *11	Poor metaboliser	23	230
<b>CYP2C9</b>	*2, *3, *4, *5, *6, *11	Poor metaboliser	24	180

\* 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	
<b>Cytochrome P450 (CYP)</b>							
<b>CYP1A2</b>	*1C	g.-3858G>A		decreased activity	25	80	
	*1E	g.-740T>G			26	80	
	*1F	g.-164C>A		higher inducibility	27	80	
	*1J	g.-740T>G g.-164C>A			28	80	
	*1K	g.-740T>G g.-730C>T g.-164C>A		decreased activity	29	80	
	*1E, *1F, *1J, *1K				30	100	
	*2	g.63C>G	F21L		31	80	
	*3	g.2116G>A	D348N		32	80	
	*4	g.2499A>G	I348N		33	80	
	*5	g.3497G>A	C406Y		34	80	
	*6	g.5090C>T	R431W		35	80	
	*7	g.3534G>A	splicing defect	decreased activity	36	80	
	<b>CYP2A6</b>	*1H	g.-745A>G			37	80
		*2	c.479T>A	L160H	no activity	38	80
*4		deletion	no protein	no activity	39	80	
*5		c.1436G>T	G479V	no activity	40	80	
*6		c.383G>A	R128Q	decreased activity	41	80	
*7		c.1412T>G	I471T	decreased activity	42	80	
*9		c.-48T>G	decreased protein	decreased activity	43	80	
*10		c.1412G>T c.1454G>T	I471T, R485L	decreased activity	44	80	

	*11	c.670T>C	S224P	decreased activity	45	80	
	*12	partial deletion	altered protein	decreased activity	46	80	
	*17	c.1093G>A	V365M	decreased activity	47	80	
	*1x2	gene duplication	increased protein	increased activity	48	80	
CYP2B6	*2	c.64C>T	R22C		49	80	
	*3	c.777C>A	S259R		50	80	
	*4	c.785A>G	K262R		51	80	
	*5	c.1459C>T	R487C		52	80	
	*6	c.516G>T c.785A>G	Q172H, K262R		53	80	
	*7	c.516G>T c.785A>G c.1459C>T	Q172H, K262R, R487C		54	80	
	*8	c.415A>G	K139E	decreased activity	55	80	
	*9	c.516G>T	Q172H		56	80	
	CYP2C8	*2	c.805A>T	I269F	increased activity	57	80
*3		c.416G>A c.1196A>G	R139K, K399R	decreased activity	58	80	
*4		c.792C>G	I264M		59	80	
*5		c.475delA	T159fs177X	no activity	60	80	
*7		c.556C>T	R186X	no activity	61	80	
*8		c.556C>G	R186G	decreased activity	62	80	
CYP2C9	*2	c.430C>T	R144C	decreased activity	63	80	
	*3	c.1075A>C	I359L	decreased activity	64	80	
	*4	c.1076T>C	I359T	decreased activity	65	80	
	*5	c.1080C>G	D360E	decreased activity	66	80	
	*6	c.818delA	frameshift	no activity	67	80	
	*7	c.55C>A	L19I		68	80	
	*8	c.449G>A	R150H	increased activity	69	80	
	*9	c.752A>G	H251R		70	80	
	*10	c.815A>G	E272G		71	80	
	*11	c.1003C>T	R335W	decreased activity	72	80	
	*12	c.1465C>T	P489S	decreased activity	73	80	
	*16	c.485C>A	T299A	decreased activity	74	80	
	*18	c.1075A>C	I359L	decreased activity	75	80	
		*2, *3, *4, *5, *6, *11			poor metaboliser	24	180
CYP2C19	*2	c.681G>A	splicing defect	no activity	76	80	
	*3	c.636G>A	W212X	no activity	77	80	
	*4	c.1A>G	start codon mutation	no activity	78	80	
	*5	c.1297G>A	R433W	no activity	79	80	
	*6	c.395G>A	R132Q	no activity	80	80	
	*7	1VS5+2T>A	splicing defect	no activity	81	80	
	*8	c.358T>C	W120R	no activity	82	80	
	*9	c.431G>A	R144H	decreased activity	83	80	
	*10	680C>T	P227L	decreased activity	84	80	
	*11	c.449G>A	R150H		85	80	
		*2, *2B, *3, *4, *5, *6, *7, *8, *9, *10, *11			poor metaboliser	23	230
	CYP2D6	*3	g.2549delA	frameshift	no activity	86	80
		*3B	g.1749A>G g.2549delA	N166D, frameshift	no activity	87	80
		*4	g.1846G>A	splicing defect	no activity	88	80
*5		gene deletion	no protein	no activity	89	80	
*6		g.1701delT	frameshift	no activity	90	80	
*7		g.2935A>C	H374P	no activity	91	80	
*8		g.1758G>T	Stop codon	no activity	92	80	
*9		g.2613-2615delAGA	K281del	decreased activity	93	80	
*14		g.1758G>A	G169R	no activity	94	80	
*17		g.1023C>T g.2850C>T	T107I, R296C	decreased activity	95	80	
*20		g.1973insG	frameshift	no activity	96	80	
*21		g.2573insC	frameshift	no activity	97	80	
*24		g.2853A>C	I297L		98	80	
*38		g.2587-2590delGACT	frameshift	no activity	99	80	
*44		g.2950G>C	splicing defect	no activity	100	80	
*XN		gene amplification	increased protein	increased activity	20	80	
		*3, *4, *5, *6, *7, *8, *9, *14, *19			poor metaboliser	19	230
		*3, *4, *5, *6, *7, *8, *9, *14, *19, *XN			poor and ultrafast metaboliser	21	270

<b>CYP2D6 / CYP2C19</b> <a href="#">Amplichip</a> <a href="#">(Roche Diagnostics)</a>	*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 *2, *3				22	900
<b>CYP2E1</b>	*1C	6 minisat			101	80
	*1D	8 minisat		increased activity	102	80
	*2	g.1132G>A	R76H	decreased activity	103	80
	*5	g.-1293G>C g.-1053C>T			104	80
	*6	g.7632T>A			105	80
	*1C, *1D				106	80
<b>CYP2J2</b>	*2	c.427A>G	T143A	decreased activity	107	80
	*3	c.472C>T	R158C	decreased activity	108	80
	*4	c.575T>A	I192N	decreased activity	109	80
	*5	c.1024G>A	D342N	decreased activity	110	80
	*6	c.1210A>T	N404Y	decreased activity	111	80
	*7	g.-76G>T	decreased protein	decreased activity	112	80
	<b>CYP3A4</b>	*1B	g.-392A>G			113
*2		g.15713T>C	S222P	decreased activity	114	80
*3		g.23172T>C	M445T		115	80
*4		g.13871A>G	I118V		116	80
*5		g.15702C>G	P218R		117	80
*6		g.17662-17663insA	frameshift		118	80
*7		g.6004G>A	G56D		119	80
*8		g.13908G>A	R130Q		120	80
*9		g.14292G>A	V170I		121	80
*10		g.14304G>C	D174H		122	80
*11		g.21867C>T	T363M		123	80
*12		g.21896C>T	L373F		124	80
*13		g.22026C>T	R416L		125	80
*14		g.44T>C	L15P		126	80
*15		g.14269G>A	R162Q		127	80
*16		g.15603C>G	T185S		128	80
*17		g.15615T>C	F189S	decreased activity	129	80
*18		g.20070T>C	L293P	increased activity	130	80
*19		g.23237C>T	P467S		131	80
<b>CYP3A5</b>	*2	g.27289C>A	T398N		132	80
	*3C	g.6986G>A	splicing defect	no activity	133	80
	*4	g.14665A>G	Q200R		134	80
	*5	g.12952T>C	splicing defect		135	80
	*6	g.14690G>A	splicing defect	no activity	136	80
	*7	g.27131-27132insT	frameshift		137	80
	*8	g.3699C>T	R28C	decreased activity	138	80
	*9	g.19386G>A	A337T	decreased activity	139	80
	*10	g.29753T>C	splicing defect	decreased activity	140	80
	<b>CYP3A7</b>	*1C	c.-291G>T -284T>A -282T>C -281A>T -270T>G -262T>A -232A>C		increased activity	141
*2		c.1226C>G	T409R	increased activity	142	80
<b>CYP4B1</b>	*2	c.881-882delAT	frameshift		143	80
<b>Epoxidhydroxylases (EPHX)</b>						
<b>EPHX1</b>	n.a.	c.128G>C	R43T		144	80
	*3	c.337T>C	Y113H		145	80
	*4	c.416A>G	H139R		146	80
<b>EPHX2</b>	n.a.	c.229A>G	K55R	increased activity	147	80
	n.a.	c.307C>T	R103C		148	80
	n.a.	c.461G>A	C154	increased activity	149	80
	n.a.	c.860G>A	R287Q	decreased activity	150	80
	n.a.	c.1208-1209insTCG	403-404insR	decreased activity	151	80
<b>Glutathione S-transferases (GST)</b>						
<b>GSTM1</b>	*0	gene deletion	no protein		152	80
<b>GSTT1</b>	*A	wild type			153	80
	*B	c.301A>C	T104P		154	80
	*0	gene deletion	no protein		155	80
<b>GSTP1</b>	*A	wild type			156	80
	*B	c.313A>G	I105V		157	80
	*C	c.313A>G c.341C>T	I105V, A114V		158	80
<b>Sulfonyl transferases (SULT)</b>						
<b>SULT1A1</b>	*2	638 G>A	R213H	decreased activity	159	80
	*3	667 G>A	M223V		160	80
	*2, *3				161	110

<b>SULT1A2</b>	*2	20 T>C	I17T		162	80	
	*3	56 T>C	P19L		163	80	
	*2, *3				164	110	
<b>N-Acetyltransferase type 2 (NAT2)</b>							
<b>NAT2</b>	*4	wild type			165	80	
	*5	c.341T>C	I114T	decreased activity	166	80	
	*6	c.590G>T	R197Q	decreased activity	167	80	
	*7	c.857G>A	G286E	decreased activity	168	80	
	*10	c.499G>A	E167K		169	80	
	*11B	c.481C>T;859del	frameshift		170	80	
	*12	c.803A>G	K268R		171	80	
	*13	c.282C>T	none		172	80	
	*14	c.191G>A	R64Q	decreased activity	173	80	
	*17	c.434A>C	Q145P	decreased activity	174	80	
	*18	c.845A>C	K282T		175	80	
	*19	c.190C>T	R64W		176	80	
		*4, *5, *6, *7, *10, *11B, *12, *13, *14, *17, *18, *19				177	200
	<b>Thiopurine methyltransferases (TPMT)</b>						
	<b>TPMT</b>	*2	c.288G>C	A80P	decreased activity	178	80
*3A		c.460G>A c.719A>G	A154T, Y240C	decreased activity	179	80	
*3B		c.719A>G	Y240C	decreased activity	180	80	
*3C		c.719A>G	A154T	decreased activity	181	80	
<b>Uridine diphosphate-glucuronyltransferases (UGT)</b>							
<b>UGT1A1</b>	*1	promoter repeat [TA] 6	wild type		182	80	
	*6	226A>G	G71R	decreased activity	183	80	
	*28	promoter repeat [TA] 7		decreased activity	184	80	
	*36	promoter repeat [TA] 5		increased activity	185	80	
	*37	promoter repeat [TA] 8		decreased activity	186	80	
		*28, **36, *37				187	80
<b>UGT1A6</b>	*1		wild type		188	80	
	*2	637A>G 648A>C	T181A, R184S	decreased activity	189	80	
<b>UGT1A7</b>	*1		wild type		190	80	
	*2	387T>G 391C>A 392G>A	N129K, R131K	decreased activity	191	80	
	*3	387T>G 391C>A 392G>A 622T>C	N129K, R131K, W208R	decreased activity	192	80	
	*4	622T>C	W208R	decreased activity	193	80	
		*1, *2, *3, *4				194	80
<b>UGT2B4</b>	*2	1411T>A	D458E	no activity	195	80	
<b>UGT2B7</b>	*2	816C>T	H268Y	decreased activity	196	80	
<b>UGT2B15</b>	*2	276T>G	D85Y	increased activity on androgens	197	80	
<b>Multidrug resistance 1 (MDR1, ABCB1)</b>							
	n.a.	1236C>T	decreased protein	decreased activity	198	80	
	n.a.	2677G>T or 2677G>A	A893S or A893T	decreased activity	199	80	
	n.a.	3435C>T	decreased protein	decreased activity	200	80	
<b>5-Hydroxytryptamine transporter (5-HTT, SLC6A4)</b>							
	L, S	44 bp insertion/deletion in the promoter region	increased protein for L (insertion) variant	increased activity for L variant	201	80	

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

**Top**

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