



New Molecular Tests (Ordered by Gene)

The molecular analysis includes analyses of all protein-coding sequences unless specified otherwise in the comment column.

If you don't find the disease (or gene) of interest in the alphabetical list, you can search the whole table with the Ctrl-F function:

1. Hold the Ctrl key, then press the F key
2. Type in the disease (gene) of interest
3. If the disease (gene) can be found, it is highlighted in black in the table

Information on mutations and genomic structure of genes can be found by clicking on [Mutation information](#).

Gene	Gene OMIM	Disease	Disease OMIM	Comment	Price in Euro
CACNA1C (CALCIUM CHANNEL, VOLTAGE-DEPENDENT, L TYPE, ALPHA-1C SUBUNIT; CACNL1A1; CCHL1A1; CaV1.2)	114205	BRUGADA SYNDROME, TYPE 3	611875		300
		TIMOTHY SYNDROME » LONG QT SYNDROME WITH SYNDACTYLY	601005		300
ERCC4 (EXCISION-REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 4; XPF)	133520	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP F, XPF » XERODERMA PIGMENTOSUM, TYPE 6, XP6	278760		1000
		XFE PROGEROID SYNDROME » XPF-ERCC1 PROGEROID SYNDROME	610965		1000
ERCC5 (EXCISION-REPAIR, COMPLEMENTING DEFECTIVE, IN CHINESE HAMSTER, 5; ERCM2, XPG, XPGC, UVDR)	133530	XERODERMA PIGMENTOSUM, COMPLEMENTATION GROUP G, XPG » XERODERMA PIGMENTOSUM, TYPE 7, XP7 » XERODERMA PIGMENTOSUM, TYPE G/COCKAYNE SYNDROME, INCLUDED » CEREBROOCULOFACIOSKELETAL SYNDROME, TYPE 3, COFS3	278780		1000
FAM126A (FAMILY WITH SEQUENCE SIMILARITY 126, MEMBER A; DOWNREGULATED BY CTNNB1, PROTEIN A; DRCTNNB1A; HYCCIN)	610531	LEUKODYSTROPHY, HYPOMYELINATING, TYPE 5 » HYPOMYELINATION AND CONGENITAL CATARACT	610532		1000
GCM2 (GLIAL CELLS MISSING, DROSOPHILA, HOMOLOG OF, 2; GCMB)	603716	HYPOPARATHYROIDISM, FAMILIAL ISOLATED » HYPOPARATHYROIDISM (AUTOSOMAL DOMINANT) » HYPOCALCEMIA (AUTOSOMAL DOMINANT) » HYPERCALCIURIC HYPOCALCEMIA, FAMILIAL	146200		480
LMNB1 (LAMIN B1)	150340	LEUKODYSTROPHY, DEMYELINATING, ADULT-ONSET (AUTOSOMAL DOMINANT), ADLD » PELIZAEUS-MERZBACHER DISEASE (AUTOSOMAL DOMINANT) OR LATE-ONSET TYPE, FORMERLY	169500		350
RNU4ATAC (RNA, U4ATAC SMALL NUCLEAR; RNA, U4, SMALL NUCLEAR, AT-AC FORM; U4ATAC)	601428	MICROCEPHALIC OSTEODYSPLASTIC PRIMORDIAL DWARFISM, TYPE 1, MOPD1 » TAYBI-LINDER SYNDROME » BRACHYMELIC PRIMORDIAL DWARFISM » CEPHALOSKELETAL DYSPLASIA	210710		480
SHOC2 (SUPPRESSOR OF CLEAR, C. ELEGANS, HOMOLOG OF; RAS-BINDING PROTEIN SUR8, C. ELEGANS, HOMOLOG OF; SUR8)	602775	NOONAN SYNDROME-LIKE DISORDER WITH LOOSE ANAGEN HAIR	607721		250

[Top](#)

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