

Gene overview

➔ Source : <http://biogps.org/#goto=genereport&id=898>
https://www.ncbi.nlm.nih.gov/protein/NP_001229.1

- Name:
- Symbole:
- Gene type: Protein -Coding Gene
- Beschreibung:
- Chromosomen:
- Expressions Ort:
- Anzahl von Proteinen:
- Function

898

- Name: cyclin E1
- Symbole: CCNE1
- Gene type: Protein Coding Gene
- Beschreibung :
- ➔ The protein encoded by this gene belongs to the highly conserved cyclin family, whose members are characterized by a dramatic periodicity in protein abundance through the cell cycle. Cyclins function as regulators of CDK kinases. Different cyclins exhibit distinct expression and degradation patterns which contribute to the temporal coordination of each mitotic event. This cyclin forms a complex with and functions as a regulatory subunit of CDK2, whose activity is required for cell cycle G1/S transition. This protein accumulates at the G1-S phase boundary and is degraded as cells progress through S phase. Overexpression of this gene has been observed in many tumors, which results in chromosome instability, and thus may contribute to tumorigenesis. This protein was found to associate with, and be involved in, the phosphorylation of NPAT protein (nuclear protein mapped to the ATM locus), which participates in cell-cycle regulated histone gene expression and plays a critical role in promoting cell-cycle progression in the absence of pRB.
[provided by RefSeq, Apr 2016]

Chromosomen: 19 (19q12)

- Expressions Ort: Placenta
- Anzahl von Proteinen: 11

Proteins:	NP_001229.1
	NP_001309188.1
	NP_001309190.1
	NP_001309191.1
	XP_011525742.1
	XP_047295562.1
	ENSP00000262643
	ENSP00000350625
	ENSP00000410179
	ENSP00000459024
	ENSP00000460891

- ➔
- Function:

	Molecular Function protein binding (GO:0005515) kinase activity (GO:0016301) cyclin-dependent protein serine/threonine kinase regulator activity (GO:0016538) protein kinase binding (GO:0019901) Biological Process regulation of cyclin-dependent protein serine/threonine kinase activity (GO:0000079) G1/S transition of mitotic cell cycle (GO:0000082) G1/S transition of mitotic cell cycle (GO:0000082) G1/S transition of mitotic cell cycle (GO:0000082) negative regulation of transcription by RNA polymerase II (GO:0001222) telomere maintenance (GO:0000723) DNA replication initiation (GO:0005279) Function: protein phosphorylation (GO:0005468) homologous chromosome pairing at meiosis (GO:0007129) Wnt signaling pathway (GO:0016055) regulation of protein localization (GO:0032880) mitotic cell cycle phase transition (GO:0044772) cell division (GO:0051301) positive regulation of mesenchymal stem cell proliferation (GO:1902462) Cellular Component cyclin-dependent protein kinase holoenzyme complex (GO:0000307) nucleus (GO:0005634) nucleus (GO:0005634) nucleoplasm (GO:0005654) nucleoplasm (GO:0005654) cytoplasm (GO:0005737) centrosome (GO:0005813) cytosol (GO:0005829) cyclin E1-CDK2 complex (GO:0097134)
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1789

- **Name: DNA methyltransferase 3 beta**
- **Symbole: DNMT3B**
- **Gene type: Protein -Coding Gene**
- **Beschreibung: CpG methylation is an epigenetic modification that is important for embryonic development, imprinting, and X-chromosome inactivation. Studies in mice have demonstrated that DNA methylation is required for mammalian development. This gene encodes a DNA methyltransferase which is thought to function in de novo methylation, rather than maintenance methylation. The protein localizes primarily to the nucleus and its expression is developmentally regulated. Mutations in this gene cause the immunodeficiency-centromeric instability-facial anomalies (ICF) syndrome. Eight alternatively spliced transcript variants have been described. The full length sequences of variants 4 and 5 have not been determined. [provided by RefSeq, May 2011]**
- **Chromosomen: 20 (20q11.21)**
- **Expressions Ort: brain,fetal and thymus**
- **Anzahl von Proteinen:**

	NP_001193984.1
	NP_001193985.1
	NP_008823.1
	NP_787044.1
	NP_787045.1
	NP_787046.1
	XP_011526955.1
	XP_011526956.1
	XP_047295902.1
	XP_047295903.1
	XP_047295904.1
	XP_047295905.1
	XP_047295906.1
	XP_047295907.1
	XP_047295908.1
	XP_047295909.1
	XP_047295910.1
	XP_047295911.1
	XP_047295912.1
	XP_047295913.1
	XP_047295914.1
	XP_047295915.1
	XP_047295916.1
	ENSP00000201963
	ENSP00000313397
	ENSP00000328547
	ENSP00000337764
	ENSP00000403169
	ENSP00000412305
	ENSP00000512497
	ENSP00000512498
	ENSP00000512499
	ENSP00000512500
	ENSP00000512501
	ENSP00000512502
	ENSP00000512503

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- **Function:**

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	Molecular Function
	DNA binding (GO:0003677)
	transcription corepressor activity (GO:0003714)
	transcription corepressor activity (GO:0003714)
	DNA (cytosine-5-)-methyltransferase activity (GO:0003886)
	DNA (cytosine-5-)-methyltransferase activity (GO:0003886)
	DNA (cytosine-5-)-methyltransferase activity (GO:0003886)
	protein binding (GO:0005515)
	DNA-methyltransferase activity (GO:0009008)
	metal ion binding (GO:0046872)
	DNA (cytosine-5-)-methyltransferase activity, acting on CpG substrates (GO:0051718)
	Biological Process
	negative regulation of transcription by RNA polymerase II (GO:0000122)
	negative regulation of transcription by RNA polymerase II (GO:0000122)
	DNA methylation (GO:0006306)
	DNA methylation (GO:0006306)
	positive regulation of gene expression (GO:0010628)
	positive regulation of histone H3-K4 methylation (GO:0051571)
	negative regulation of histone H3-K9 methylation (GO:0051573)
	C-5 methylation of cytosine (GO:0090116)
	C-5 methylation of cytosine (GO:0090116)
	Cellular Component
	nucleus (GO:0005634)
	nucleus (GO:0005634)
	nucleus (GO:0005634)
	nucleoplasm (GO:0005654)
	nucleoplasm (GO:0005654)
	catalytic complex (GO:1902494)

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26227

- **Name: phosphoglycerate dehydrogenase**

- **Symbole:** PHGDH
- **Gene type:** Protein -Coding Gene
- **Beschreibung:** This gene encodes the enzyme which is involved in the early steps of L-serine synthesis in animal cells. L-serine is required for D-serine and other amino acid synthesis. The enzyme requires NAD/NADH as a cofactor and forms homotetramers for activity. Mutations in this gene have been found in a family with congenital microcephaly, psychomotor retardation and other symptoms. Multiple alternatively spliced transcript variants have been found, however the full-length nature of most are not known. [provided by RefSeq, Aug 2011]
- **Chromosomen:** 1 (1p12)
- **Expressions Ort:** Prostate ,brain Whole and Salivary gland
- **Anzahl von Proteinen:**

Proteins:	NP_001193984.1
	NP_001193985.1
	NP_008823.1
	NP_787044.1
	NP_787045.1
	NP_787046.1
	XP_011526955.1
	XP_011526956.1
	XP_047295902.1
	XP_047295903.1
	XP_047295904.1
	XP_047295905.1
	XP_047295906.1
	XP_047295907.1
	XP_047295908.1
	XP_047295909.1
	XP_047295910.1
	XP_047295911.1
	XP_047295912.1
	XP_047295913.1
	XP_047295914.1
	XP_047295915.1
	XP_047295916.1
	ENSP00000201963
	ENSP00000313397
	ENSP00000328547
	ENSP00000337764
	ENSP00000403169
	ENSP00000412305
	ENSP00000512497
	ENSP00000512498
	ENSP00000512499
	ENSP00000512500
	ENSP00000512501
	ENSP00000512502
	ENSP00000512503

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- **Function**

Function:	Molecular Function
	DNA binding (GO:0003677)
	transcription corepressor activity (GO:0003714)
	transcription corepressor activity (GO:0003714)
	DNA (cytosine-5-)-methyltransferase activity (GO:0003886)
	DNA (cytosine-5-)-methyltransferase activity (GO:0003886)
	DNA (cytosine-5-)-methyltransferase activity (GO:0003886)
	protein binding (GO:0005515)
	DNA-methyltransferase activity (GO:0009008)
	metal ion binding (GO:0046872)
	DNA (cytosine-5-)-methyltransferase activity, acting on CpG substrates (GO:0051718)
	Biological Process
	negative regulation of transcription by RNA polymerase II (GO:0000122)
	negative regulation of transcription by RNA polymerase II (GO:0000122)
	DNA methylation (GO:0006306)
	DNA methylation (GO:0006306)
	positive regulation of gene expression (GO:0010628)
	positive regulation of histone H3-K4 methylation (GO:0051571)
	negative regulation of histone H3-K9 methylation (GO:0051573)
	C-5 methylation of cytosine (GO:0090116)
	C-5 methylation of cytosine (GO:0090116)
	Cellular Component
	nucleus (GO:0005634)
	nucleus (GO:0005634)
	nucleus (GO:0005634)
	nucleoplasm (GO:0005654)
	nucleoplasm (GO:0005654)
	catalytic complex (GO:1902494)

2886

- **Name:** growth factor receptor bound protein 7
- **Symbole:** GRB7
- **Gene type:** Protein -Coding Gene
- **Beschreibung:** The product of this gene belongs to a small family of adapter proteins that are known to interact with a number of receptor tyrosine kinases and signaling molecules. This gene encodes a growth factor receptor-binding protein that interacts with epidermal growth factor receptor (EGFR) and ephrin receptors. The protein plays a role in the integrin signaling pathway and cell migration by binding with focal adhesion kinase (FAK). Several transcript variants encoding two different isoforms have been found for this gene. [provided by RefSeq, Jun 2011]
- **Chromosomen:** 17(17q12)
- **Expressions Ort:** Placenta
- **Anzahl von Proteinen:**

Proteins:	NP_001025173.1
	NP_001229371.2
	NP_001229372.1
	NP_001317136.1
	NP_005301.2
	XP_047291813.1
	ENSP00000310771
	ENSP00000377754
	ENSP00000377759
	ENSP00000377761
	ENSP00000403459
	ENSP00000463126
	ENSP00000463416

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- **Function**

Function:	Molecular Function
	RNA binding (GO:0003723)
	protein binding (GO:0005515)
	protein kinase binding (GO:0019901)
	phosphatidylinositol binding (GO:0035091)
	identical protein binding (GO:0042802)
	Biological Process
	epidermal growth factor receptor signaling pathway (GO:0007173)
	insulin receptor signaling pathway (GO:0008286)
	negative regulation of translation (GO:0017148)
	positive regulation of cell migration (GO:0030335)
	stress granule assembly (GO:0034063)
	negative regulation of insulin receptor signaling pathway (GO:0046627)
	Cellular Component
	cytosol (GO:0005829)
	cytosol (GO:0005829)
	plasma membrane (GO:0005886)
	focal adhesion (GO:0005925)
	cytoplasmic stress granule (GO:0010494)
	cell projection (GO:0042995)

8648

- **Name:** nuclear receptor coactivator 1
- **Symbole:** NCOA1
- **Gene type:** Protein -Coding Gene
- **Beschreibung:** The protein encoded by this gene acts as a transcriptional coactivator for steroid and nuclear hormone receptors. It is a member of the p160/steroid receptor coactivator (SRC) family and like other family members has histone acetyltransferase activity and contains a nuclear localization signal, as well as bHLH and PAS domains. The product of this gene binds nuclear receptors directly and stimulates the transcriptional activities in a hormone-

dependent fashion. Alternatively spliced transcript variants encoding different isoforms have been identified. [provided by RefSeq, Jul 2008]

- Chromosomen: 2(2p23.3)
- Expressions Ort: brain cerebellum
- Anzahl von Proteinen:

Proteins:	NP_001349879.1
	NP_001349881.1
	NP_001349883.1
	NP_001349884.1
	NP_003734.3
	NP_671756.1
	NP_671766.1
	XP_047302107.1
	XP_047302108.1
	XP_047302109.1
	XP_047302110.1
	XP_047302111.1
	XP_047302112.1
	XP_047302113.1
	XP_047302114.1
	ENSP00000288599
	ENSP00000320940
	ENSP00000379197
	ENSP00000385097
	ENSP00000385195
	ENSP00000385216

- Function

Function:	Molecular Function DNA binding (GO:0003677) chromatin binding (GO:0003682) transcription coactivator activity (GO:0003713) transcription coactivator activity (GO:0003713) histone acetyltransferase activity (GO:0004402) protein binding (GO:0005515) nuclear receptor binding (GO:0016922) nuclear receptor binding (GO:0016922) nuclear receptor binding (GO:0016922) aryl hydrocarbon receptor binding (GO:0017162) enzyme binding (GO:0019899) nuclear estrogen receptor binding (GO:0030331) nuclear receptor coactivator activity (GO:0030374) nuclear receptor coactivator activity (GO:0030374) protein dimerization activity (GO:0046983) protein N-terminus binding (GO:0047485) Biological Process positive regulation of transcription from RNA polymerase II promoter by galactose (GO:0000435) regulation of thyroid hormone mediated signaling pathway (GO:0002155) cellular response to hormone stimulus (GO:0032870) peroxisome proliferator activated receptor signaling pathway (GO:0035357) mRNA transcription by RNA polymerase II (GO:0042789) positive regulation of apoptotic process (GO:0043065) histone H4 acetylation (GO:0043967)	Function: (GO:0035357) mRNA transcription by RNA polymerase II (GO:0042789) positive regulation of apoptotic process (GO:0043065) histone H4 acetylation (GO:0043967) positive regulation of neuron differentiation (GO:0045666) positive regulation of DNA-templated transcription (GO:0045893) positive regulation of DNA-templated transcription (GO:0045893) positive regulation of transcription by RNA polymerase II (GO:0045944) positive regulation of transcription by RNA polymerase II (GO:0045944) positive regulation of transcription by RNA polymerase II (GO:0045944) labyrinthine layer morphogenesis (GO:0060713) regulation of cellular response to insulin stimulus (GO:1900076) cellular response to Thyroglobulin triiodothyronine (GO:1904017) positive regulation of adipose tissue development (GO:1904179) Cellular Component chromatin (GO:0000785) chromatin (GO:0000785) nucleus (GO:0005634) nucleoplasm (GO:0005654) nucleoplasm (GO:0005654) transcription regulator complex (GO:0005667) cytosol (GO:0005829) plasma membrane (GO:0005886) protein-containing complex (GO:0032991) RNA polymerase II transcription regulator complex (GO:0090575)
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2690

- Name: growth hormone receptor
- Symbole: GHR
- Gene type: Protein -Coding Gene

- **Beschreibung:** This gene encodes a member of the type I cytokine receptor family, which is a transmembrane receptor for growth hormone. Binding of growth hormone to the receptor leads to receptor dimerization and the activation of an intra- and intercellular signal transduction pathway leading to growth. Mutations in this gene have been associated with Laron syndrome, also known as the growth hormone insensitivity syndrome (GHIS), a disorder characterized by short stature. In humans and rabbits, but not rodents, growth hormone binding protein (GHBP) is generated by proteolytic cleavage of the extracellular ligand-binding domain from the mature growth hormone receptor protein. Multiple alternatively spliced transcript variants have been found for this gene.[provided by RefSeq, Jun 2011]

Chromosomen: 5(5p13.1-p12)

- **Expressions Ort:** Liver

- **Anzahl von Proteinen:**

Proteins:	NP_000154.1
	NP_001229328.1
	NP_001229329.1
	NP_001229330.1
	NP_001229331.1
	NP_001229332.1
	NP_001229333.1
	NP_001229334.1
	NP_001229335.1
	NP_001229389.1
	NP_001229391.1
	ENSP00000230882
	ENSP00000350335
	ENSP00000422333
	ENSP00000426739
	ENSP00000442206
	ENSP00000478291
	ENSP00000478332
	ENSP00000479846
	ENSP00000482373
	ENSP00000483403
	ENSP00000483926

- **Function:**

<p>Molecular Function</p> <p>cytokine receptor activity (GO:0004896)</p> <p>growth hormone receptor activity (GO:0004903)</p> <p>protein binding (GO:0005515)</p> <p>peptide hormone binding (GO:0017046)</p> <p>peptide hormone binding (GO:0017046)</p> <p>growth factor binding (GO:0019838)</p> <p>protein kinase binding (GO:0019901)</p> <p>protein phosphatase binding (GO:0019903)</p> <p>cytokine binding (GO:0019955)</p> <p>SH2 domain binding (GO:0042169)</p> <p>identical protein binding (GO:0042802)</p> <p>protein homodimerization activity (GO:0042803)</p> <p>proline-rich region binding (GO:0070064)</p> <p>Biological Process</p> <p>endocytosis (GO:0006897)</p> <p>receptor signaling pathway via JAK-STAT (GO:0007259)</p> <p>response to gravity (GO:0009629)</p> <p>hormone-mediated signaling pathway (GO:0009755)</p> <p>cytokine-mediated signaling pathway (GO:0019221)</p> <p>taurine metabolic process (GO:0019530)</p> <p>response to food (GO:0032094)</p> <p>cellular response to insulin stimulus (GO:0032869)</p> <p>cellular response to hormone stimulus (GO:0032870)</p> <p>regulation of multicellular organism growth (GO:0040014)</p> <p>positive regulation of multicellular organism growth (GO:0040018)</p> <p>hormone metabolic process (GO:0042445)</p> <p>positive regulation of tyrosine phosphorylation of STAT protein (GO:0042531)</p>	<p>Function:</p> <p>activation of Janus kinase activity (GO:0042976)</p> <p>response to morphine (GO:0043278)</p> <p>positive regulation of MAP kinase activity (GO:0043406)</p> <p>positive regulation of cell differentiation (GO:0045597)</p> <p>positive regulation of receptor signaling pathway via JAK-STAT (GO:0046427)</p> <p>insulin-like growth factor receptor signaling pathway (GO:0048009)</p> <p>positive regulation of peptidyl-tyrosine phosphorylation (GO:0050731)</p> <p>positive regulation of peptidyl-tyrosine phosphorylation (GO:0050731)</p> <p>response to glucocorticoid (GO:0051384)</p> <p>cartilage development involved in endochondral bone morphogenesis (GO:0060351)</p> <p>growth hormone receptor signaling pathway (GO:0060396)</p> <p>growth hormone receptor signaling pathway (GO:0060396)</p> <p>response to interleukin-1 (GO:0070555)</p> <p>negative regulation of neuron death (GO:1901215)</p> <p>Cellular Component</p> <p>extracellular region (GO:0005576)</p> <p>extracellular space (GO:0005615)</p> <p>extracellular space (GO:0005615)</p> <p>nucleus (GO:0005634)</p> <p>mitochondrion (GO:0005739)</p> <p>cytosol (GO:0005829)</p> <p>cytosol (GO:0005829)</p> <p>plasma membrane (GO:0005886)</p> <p>plasma membrane (GO:0005886)</p> <p>integral component of plasma membrane (GO:0005887)</p> <p>external side of plasma membrane (GO:0009897)</p> <p>cell surface (GO:0009986)</p> <p>integral component of membrane (GO:0016021)</p> <p>cytoplasmic ribonucleoprotein granule</p>	<p>(GO:0016021)</p> <p>cytoplasmic ribonucleoprotein granule (GO:0036464)</p> <p>neuronal cell body (GO:0043025)</p> <p>receptor complex (GO:0043235)</p> <p>receptor complex (GO:0043235)</p> <p>growth hormone receptor complex (GO:0070195)</p> <p>growth hormone receptor complex (GO:0070195)</p>
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4319

- **Name:** matrix metalloproteinase 10
- **Symbole:** MMP10
- **Gene type:** Protein -Coding Gene
- **Beschreibung:** This gene encodes a member of the peptidase M10 family of matrix metalloproteinases (MMPs). Proteins in this family are involved in the breakdown of extracellular matrix in normal physiological processes, such as embryonic development, reproduction, and tissue remodeling, as well as in disease processes, such as arthritis and metastasis. The encoded preproprotein is proteolytically processed to generate the mature protease. This secreted protease breaks down fibronectin, laminin, elastin, proteoglycan core protein, gelatins, and several types of collagen. The gene is part of a cluster of MMP genes on chromosome 11. [provided by RefSeq, Jan 2016]
- **Chromosomen:** 11(11q22.2)
- **Expressions Ort:** Uterus and trachea
- **Anzahl von Proteinen:**
- **Proteins:** NP_002416.1
ENSP00000279441
ENSP00000441485
- **Function**

Function:	Molecular Function
	metalloendopeptidase activity
	(GO:0004222)
	metalloendopeptidase activity
	(GO:0004222)
	serine-type endopeptidase activity
	(GO:0004252)
	zinc ion binding (GO:0008270)
	Biological Process
	proteolysis (GO:0006508)
	extracellular matrix disassembly
	(GO:0022617)
	extracellular matrix organization
	(GO:0030198)
	collagen catabolic process
	(GO:0030574)
	collagen catabolic process
	(GO:0030574)
	Cellular Component
	extracellular region (GO:0005576)
	extracellular space (GO:0005615)
	extracellular matrix (GO:0031012)

7170

- **Name: tropomyosin 3**
- **Symbole: TPM3**
- **Gene type: Protein -Coding Gene**
- **Beschreibung:** This gene encodes a member of the tropomyosin family of actin-binding proteins. Tropomyosins are dimers of coiled-coil proteins that provide stability to actin filaments and regulate access of other actin-binding proteins. Mutations in this gene result in autosomal dominant nemaline myopathy and other muscle disorders. This locus is involved in translocations with other loci, including anaplastic lymphoma receptor tyrosine kinase (ALK) and neurotrophic tyrosine kinase receptor type 1 (NTRK1), which result in the formation of fusion proteins that act as oncogenes. There are numerous pseudogenes for this gene on different chromosomes. Alternative splicing results in multiple transcript variants. [provided by RefSeq, May 2013]
Chromosomen: 16(1q21.3)
- **Expressions Ort: skeletal muscle**
- **Anzahl von Proteinen:**

Proteins:	NP_001036816.1
	NP_001036817.1
	NP_001036818.1
	NP_001265117.1
	NP_001265118.1
	NP_001265119.1
	NP_001265120.1
	NP_001336608.1
	NP_001351608.1
	NP_001351609.1
	NP_001351610.1
	NP_001351611.1
	NP_001351612.1
	NP_689476.2
	NP_705935.1
	ENSP00000271850
	ENSP00000307712
	ENSP00000339035
	ENSP00000339378
	ENSP00000341653
	ENSP00000357516
	ENSP00000357517
	ENSP00000357518
	ENSP00000357520
	ENSP00000357521
	ENSP00000422207
	ENSP00000426306
	ENSP00000426521
	ENSP00000480520
	ENSP00000498347
	ENSP00000498531
	ENSP00000498577
	ENSP00000498648

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- **Function**

Function:	Molecular Function
	molecular_function (GO:0003674)
	protein binding (GO:0005515)
	actin filament binding (GO:0051015)
	Biological Process
	muscle contraction (GO:0006936)
	actin filament organization (GO:0007015)
	Cellular Component
	stress fiber (GO:0001725)
	cytosol (GO:0005829)
	cytosol (GO:0005829)
	cytoskeleton (GO:0005856)
	cytoskeleton (GO:0005856)
	muscle thin filament tropomyosin (GO:0005862)
	actin filament (GO:0005884)
	actin cytoskeleton (GO:0015629)
	extracellular exosome (GO:0070062)

2180

- **Name:** acyl-CoA synthetase long chain family member 1
- **Symbole:** ACSL1
- **Gene type:** Protein -Coding Gene
- **Beschreibung:** The protein encoded by this gene is an isozyme of the long-chain fatty-acid-coenzyme A ligase family. Although differing in substrate specificity, subcellular localization, and tissue distribution, all isozymes of this family convert free long-chain fatty acids into fatty acyl-CoA esters, and thereby play a key role in lipid biosynthesis and fatty acid degradation. Several transcript variants encoding different isoforms have been found for this gene. [provided by RefSeq, Nov 2013]
- **Chromosomen:** 4(4q35.1)
- **Expressions Ort:** Liver and Kidney
- **Anzahl von Proteinen:**

Proteins:	NP_001273637.1
	NP_001273639.1
	NP_001273640.1
	NP_001368806.1
	NP_001368807.1
	NP_001368808.1
	NP_001368809.1
	NP_001368810.1
	NP_001368811.1
	NP_001368812.1
	NP_001368813.1
	NP_001368814.1
	NP_001368815.1
	NP_001368816.1
	NP_001368817.1
	NP_001368818.1
	NP_001368819.1
	NP_001986.2
	XP_016863376.1
	XP_047305775.1
	ENSP00000281455
	ENSP00000407165
	ENSP00000422607
	ENSP00000424935
	ENSP00000425006
	ENSP00000425098
	ENSP00000425640
	ENSP00000426150
	ENSP00000426244
	ENSP00000426491

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- **Function**

Function:	Molecular Function long-chain fatty acid-CoA ligase activity (GO:0004467) long-chain fatty acid-CoA ligase activity (GO:0004467) long-chain fatty acid-CoA ligase activity (GO:0004467) ATP binding (GO:0005524) arachidonate-CoA ligase activity (GO:0047676) arachidonate-CoA ligase activity (GO:0047676) phytanate-CoA ligase activity (GO:0050197) pristanate-CoA ligase activity (GO:0070251) oleoyl-CoA ligase activity (GO:0090434) Biological Process very long-chain fatty acid metabolic process (GO:0000038) long-chain fatty acid metabolic process (GO:0001676) response to nutrient (GO:0007584) lipid biosynthetic process (GO:0008610) lipid biosynthetic process (GO:0008610) lipid biosynthetic process (GO:0008610) positive regulation of long-chain fatty acid import across plasma membrane (GO:0010747) response to organic cyclic compound (GO:0014070) fatty acid transport (GO:0015908) triglyceride biosynthetic process (GO:0019432) adiponectin-activated signaling pathway (GO:0033211) response to oleic acid (GO:0034201) long-chain fatty-acyl-CoA biosynthetic process (GO:0035338) long-chain fatty-acyl-CoA biosynthetic	long-chain fatty-acyl-CoA biosynthetic process (GO:0035338) long-chain fatty-acyl-CoA biosynthetic process (GO:0035338) xenobiotic catabolic process (GO:0042178) long-chain fatty acid import into cell (GO:0044539) long-chain fatty acid import into cell (GO:0044539) positive regulation of protein serine/threonine kinase activity (GO:0071902) positive regulation of cold-induced thermogenesis (GO:0120162) Cellular Component mitochondrion (GO:0005739) mitochondrion (GO:0005739) mitochondrial outer membrane (GO:0005741) peroxisomal membrane (GO:0005778) endoplasmic reticulum (GO:0005783) endoplasmic reticulum (GO:0005783) endoplasmic reticulum membrane (GO:0005789) plasma membrane (GO:0005886) membrane (GO:0016020) membrane (GO:0016020) integral component of membrane (GO:0016021)
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5618

- **Name:** prolactin receptor
- **Symbole:** PRLR
- **Gene type:** Protein -Coding Gene
- **Beschreibung:** This gene encodes a receptor for the anterior pituitary hormone, prolactin, and belongs to the type I cytokine receptor family. Prolactin-dependent signaling occurs as the result of ligand-induced dimerization of the prolactin receptor. Several alternatively spliced transcript variants encoding different membrane-bound and soluble isoforms have been described for this gene, which may function to modulate the endocrine and autocrine effects of prolactin in normal tissue and cancer. [provided by RefSeq, Feb 2011]
- **Chromosomen:** 5(5p13.2)
- **Expressions Ort:** Placenta
- **Anzahl von Proteinen:**

	NP_000940.1
	NP_001191243.1
	NP_001191244.1
	NP_001191245.1
	NP_001191246.1
	NP_001191247.1
	XP_006714547.1
	XP_011512370.1
	XP_024301899.1
	XP_047273344.1
	XP_047273346.1
	XP_047273347.1
	ENSP00000231423
	ENSP00000309008
	ENSP00000311613
Proteins:	ENSP00000380546
	ENSP00000421864
	ENSP00000422385
	ENSP00000422556
	ENSP00000422867
	ENSP00000422935
	ENSP00000423493
	ENSP00000424841
	ENSP00000425300
	ENSP00000427060
	ENSP00000427236
	ENSP00000441813
	ENSP00000479077
	ENSP00000482689
	ENSP00000482954
	ENSP00000484768

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- **Function**

	Molecular Function cytokine receptor activity (GO:0004896) leukemia inhibitory factor receptor activity (GO:0004923) oncostatin-M receptor activity (GO:0004924) prolactin receptor activity (GO:0004925) prolactin receptor activity (GO:0004925) ciliary neurotrophic factor receptor binding (GO:0005127) protein binding (GO:0005515) peptide hormone binding (GO:0017046) peptide hormone binding (GO:0017046) protein kinase binding (GO:0019901) cytokine binding (GO:0019955) metal ion binding (GO:0046872) Biological Process steroid biosynthetic process (GO:0006694) activation of transmembrane receptor protein tyrosine kinase activity (GO:0007171) receptor signaling pathway via JAK-STAT (GO:0007259) embryo implantation (GO:0007566) lactation (GO:0007595) positive regulation of cell population proliferation (GO:0008284) response to bacterium (GO:0009617) cytokine-mediated signaling pathway (GO:0019221) regulation of cell adhesion (GO:0030155) regulation of epithelial cell differentiation (GO:0030856) prolactin signaling pathway (GO:0038161) oncostatin-M-mediated signaling pathway (GO:0038165) activation of Janus kinase activity (GO:0042976)	negative regulation of apoptotic process (GO:0043066) leukemia inhibitory factor signaling pathway (GO:0048861) mammary gland epithelial cell differentiation (GO:0060644) prostate gland growth (GO:0060736) mammary gland alveolus development (GO:0060749) positive regulation of cold-induced thermogenesis (GO:0120162) Cellular Component extracellular region (GO:0005576) plasma membrane (GO:0005886) external side of plasma membrane (GO:0009897) cell surface (GO:0009986) integral component of membrane (GO:0016021) endosome lumen (GO:0031904) receptor complex (GO:0043235)
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4321

- **Name:** matrix metalloproteinase 12
- **Symbole:** MMP12
- **Gene type:** Protein -Coding Gene
- **Beschreibung:** This gene encodes a member of the peptidase M10 family of matrix metalloproteinases (MMPs). Proteins in this family are involved in the

breakdown of extracellular matrix in normal physiological processes, such as embryonic development, reproduction, and tissue remodeling, as well as in disease processes, such as arthritis and metastasis. The encoded preproprotein is proteolytically processed to generate the mature protease. This protease degrades soluble and insoluble elastin. This gene may play a role in aneurysm formation and mutations in this gene are associated with lung function and chronic obstructive pulmonary disease (COPD). This gene is part of a cluster of MMP genes on chromosome 11. [provided by RefSeq, Jan 2016]

- Chromosomen: 10(11q22.2)

- Expressions Ort: Stomach

- Anzahl von Proteinen:

-> Proteins: [NP_002417.2](#)
[ENSP00000458585](#)

- Function

Molecular Function	Function:
core promoter sequence-specific DNA binding (GO:0001046)	positive regulation of interferon-alpha production (GO:0032727)
endopeptidase activity (GO:0004175)	wound healing, spreading of epidermal cells (GO:0035313)
metalloendopeptidase activity (GO:0004222)	positive regulation of transcription by RNA polymerase II (GO:0045944)
metalloendopeptidase activity (GO:0004222)	lung alveolus development (GO:0048286)
metalloendopeptidase activity (GO:0004222)	regulation of defense response to virus by host (GO:0050691)
metalloendopeptidase activity (GO:0004222)	positive regulation of epithelial cell proliferation involved in wound healing (GO:0060054)
serine-type endopeptidase activity (GO:0004252)	elastin catabolic process (GO:0060309)
calcium ion binding (GO:0005509)	negative regulation of type I interferon-mediated signaling pathway (GO:0060339)
collagen binding (GO:0005518)	positive regulation of type I interferon-mediated signaling pathway (GO:0060340)
zinc ion binding (GO:0008270)	bronchiole development (GO:0060435)
sequence-specific DNA binding (GO:0043565)	cellular response to virus (GO:0098586)
Biological Process	response to amyloid-beta (GO:1904645)
negative regulation of transcription by RNA polymerase II (GO:0000122)	negative regulation of endothelial cell-matrix adhesion via fibronectin (GO:1904905)
proteolysis (GO:0006508)	Cellular Component
proteolysis (GO:0006508)	extracellular region (GO:0005576)
protein import into nucleus (GO:0006606)	extracellular space (GO:0005615)
extracellular matrix disassembly (GO:0022617)	nucleus (GO:0005634)
extracellular matrix organization (GO:0030198)	cytoplasm (GO:0005737)
collagen catabolic process (GO:0030574)	extracellular matrix (GO:0031012)
collagen catabolic process (GO:0030574)	

10125

- Name: RAS guanyl releasing protein 1

- Symbole: RASGRP1

- Gene type: Protein -Coding Gene

- Beschreibung: This gene is a member of a family of genes characterized by the presence of a Ras superfamily guanine nucleotide exchange factor (GEF) domain. It functions as a diacylglycerol (DAG)-regulated nucleotide exchange factor specifically activating Ras through the exchange of bound GDP for GTP. It activates the Erk/MAP kinase cascade and regulates T-cells and B-cells development, homeostasis and differentiation. Alternatively spliced transcript variants encoding different isoforms have been identified. Altered expression of the different isoforms of this protein may be a cause of susceptibility to systemic lupus erythematosus (SLE). [provided by RefSeq, Jul 2008]

Chromosomen: 15(15q14)

- Expressions Ort: thymus,(breain,cerebellum)

- Anzahl von Proteinen:

	NP_001122074.1
	NP_001293015.1
	NP_005730.2
	XP_047288029.1
	XP_047288030.1
	XP_047288031.1
	XP_047288032.1
	XP_047288033.1
	XP_047288034.1
	ENSP00000310244
Proteins:	ENSP00000388540
	ENSP00000413105
	ENSP00000452721
	ENSP00000452859
	ENSP00000452892
	ENSP00000453583
	ENSP00000453729
	ENSP00000453905
	ENSP00000454005
	ENSP00000454053
	ENSP00000454164

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- Function

	Molecular Function guanyl-nucleotide exchange factor activity (GO:0005085) guanyl-nucleotide exchange factor activity (GO:0005085) guanyl-nucleotide exchange factor activity (GO:0005085) calcium ion binding (GO:0005509) zinc ion binding (GO:0008270) lipid binding (GO:0008289) diacylglycerol binding (GO:0019992) phosphatidylcholine binding (GO:0031210) identical protein binding (GO:0042802) Biological Process positive regulation of protein phosphorylation (GO:0001934) positive regulation of protein phosphorylation (GO:0001934) inflammatory response to antigenic stimulus (GO:0002437) signal transduction (GO:0007165) Ras protein signal transduction (GO:0007265) Ras protein signal transduction (GO:0007265) regulation of phosphatidylinositol 3-kinase signaling (GO:0014066) natural killer cell activation (GO:0030101) cell differentiation (GO:0030154) secretory granule localization (GO:0032252) positive regulation of granulocyte macrophage colony-stimulating factor production (GO:0032725) positive regulation of interferon-gamma production (GO:0032729) positive regulation of tumor necrosis factor production (GO:0032760) positive regulation of natural killer cell differentiation (GO:0032825) positive regulation of T cell differentiation in thymus (GO:0033089) T cell proliferation (GO:0042098)	Function: positive regulation of T cell differentiation in thymus (GO:0033089) T cell proliferation (GO:0042098) B cell proliferation (GO:0042100) T cell activation (GO:0042110) B cell activation (GO:0042113) mast cell degranulation (GO:0043303) positive regulation of MAP kinase activity (GO:0043406) positive regulation of GTPase activity (GO:0043547) positive regulation of GTPase activity (GO:0043547) positive regulation of natural killer cell mediated cytotoxicity (GO:0045954) positive regulation of JNK cascade (GO:0046330) positive regulation of Ras protein signal transduction (GO:0046579) vesicle transport along microtubule (GO:0047496) regulation of ERK1 and ERK2 cascade (GO:0070372) positive regulation of ERK1 and ERK2 cascade (GO:0070374) activation of GTPase activity (GO:0090630) activation of GTPase activity (GO:0090630) regulation of phospholipase C activity (GO:1900274) regulation of p38MAPK cascade (GO:1900744) Cellular Component Golgi membrane (GO:0000139) endoplasmic reticulum membrane (GO:0005789) Golgi apparatus (GO:0005794) cytosol (GO:0005829) cytosol (GO:0005829) plasma membrane (GO:0005886) plasma membrane (GO:0005886) plasma membrane (GO:0005886) membrane (GO:0016020)
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