Oslo universitetssykehus
Avdeling for medisinsk genetikk

Genpanel for monogen diabetes mellitus

Genpanel, versjon v01

* Enkelte genomiske regioner har lav eller ingen sekvensdekning ved eksomsekvensering. Dette skyldes at de har stor likhet med andre områder i genomet, slik at spesifikk gjenkjennelse av disse områdene og påvisning av varianter i disse områdene, blir vanskelig og upålitelig. Disse genetiske regionene har vi identifisert ved å benytte USCS segmental duplication hvor områder større enn 1 kb og ≥90% likhet med andre regioner i genomet, gjenkjennes (https://genome.ucsc.edu).

Vi gjør oppmerksom på at ved identifiseringav ekson oppstrøms for startkodon kan eksonnummereringen endres uten at transkript ID endres.

Avdelingens websider har en full oversikt over områder som er affisert av segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
ABCC8	59	NM_000352.4		1-39	Diabetes mellitus, noninsulin-dependent OMIM Diabetes mellitus, permanent neonatal 3, with or without neurologic features OMIM Diabetes mellitus, transient neonatal 2 OMIM Hyperinsulinemic hypoglycemia, familial, 1 OMIM Hypoglycemia of infancy, leucine-sensitive OMIM
AGPAT2	325	NM_006412.4		1-6	Lipodystrophy, congenital generalized, type 1 OMIM
AKT2	392	NM_001626.5		2-14	Diabetes mellitus, type II OMIM Hypoinsulinemic hypoglycemia with hemihypertrophy OMIM
ALMS1	428	NM_015120.4	17-21	1-23	Alstrom syndrome OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson affisert av segdup*	Ekson**	Fenotype
APPL1	24035	NM_012096.2		1-22	{Maturity-onset diabetes of the young, type 14} OMIM
BSCL2	15832	NM_001122955.3		1-11	Encephalopathy, progressive, with or without lipodystrophy OMIM Lipodystrophy, congenital generalized, type 2 OMIM Neuropathy, distal hereditary motor, type VC OMIM Silver spastic paraplegia syndrome OMIM
CISD2	24212	NM_001008388.4	3	1-3	Wolfram syndrome 2 OMIM
DCAF17	25784	NM_025000.3		1-14	Woodhouse-Sakati syndrome OMIM
DMXL2	2938	NM_015263.4		1-43	Developmental and epileptic encephalopathy 81 OMIM
DYRK1B	3092	NM_004714.2		2-11	Abdominal obesity-metabolic syndrome 3 OMIM
EIF2AK3	3255	NM_004836.5		1-17	Wolcott-Rallison syndrome OMIM
EIF2S3	3267	NM_001415.3	12	1-12	MEHMO syndrome OMIM
FOXP3	6106	NM_014009.3		2-12	Immunodysregulation, polyendocrinopathy, and enteropathy, X-linked OMIM
GCK	4195	NM_000162.3		1-10	Diabetes mellitus, noninsulin-dependent, late onset OMIM Diabetes mellitus, permanent neonatal 1 OMIM Hyperinsulinemic hypoglycemia, familial, 3 OMIM MODY, type II OMIM
GLIS3	28510	NM_001042413.1		2-11	Diabetes mellitus, neonatal, with congenital hypothyroidism OMIM
HAMP	15598	NM_021175.3		1-3	Hemochromatosis, type 2B OMIM

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HFE	4886	NM_000410.3		1-6	Hemochromatosis OMIM
HNF1A	11621	NM_000545.5		1-10	Diabetes mellitus, insulin-dependent, 20 OMIM Hepatic adenoma, somatic OMIM MODY, type III OMIM
HNF1B	11630	NM_001165923.3		1-9	Type 2 diabetes mellitus OMIM
HNF4A	5024	NM_175914.4		1-10	Fanconi renotubular syndrome 4, with maturity-onset diabetes of the young OMIM MODY, type I OMIM {Diabetes mellitus, noninsulin-dependent} OMIM
IER3IP1	18550	NM_016097.4		1-3	Microcephaly, epilepsy, and diabetes syndrome OMIM
IL2RA	6008	NM_000417.3		1-8	{Diabetes, mellitus, insulin-dependent, susceptibility to, 10} OMIM
INS	6081	NM_001185097.2		2-3	Diabetes mellitus, insulin-dependent, 2 OMIM Diabetes mellitus, permanent neonatal 4 OMIM Maturity-onset diabetes of the young, type 10 OMIM
INSR	6091	NM_001079817.1		1-21	Diabetes mellitus, insulin-resistant, with acanthosis nigricans OMIM Hyperinsulinemic hypoglycemia, familial, 5 OMIM
KCNJ11	6257	NM_001166290.1		2	Diabetes mellitus, transient neonatal 3 OMIM Diabetes, permanent neonatal 2, with or without neurologic features OMIM Hyperinsulinemic hypoglycemia, familial, 2 OMIM Maturity-onset diabetes of the young, type 13 OMIM

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KLF11	11811	NM_003597.4		1-4	Maturity-onset diabetes of the young, type VII OMIM
LMNA	6636	NM_001282625.1		4-13	Lipodystrophy, familial partial, type 2 OMIM
LRBA	1742	NM_006726.4		2-58	Immunodeficiency, common variable, 8, with autoimmunity OMIM
MNX1	4979	NM_005515.3		1-3	Currarino syndrome OMIM
NEUROD1	7762	NM_002500.4		2	Maturity-onset diabetes of the young 6 OMIM
PAX4	8618	NM_006193.2		1-9	Diabetes mellitus, type 2 OMIM Maturity-onset diabetes of the young, type IX OMIM
PCBD1	8646	NM_000281.3		1-4	Hyperphenylalaninemia, BH4-deficient, D OMIM
PDX1	6107	NM_000209.3		1-2	MODY, type IV OMIM
PIK3R1	8979	NM_181523.3		2-16	SHORT syndrome OMIM
PLIN1	9076	NM_001145311.1		2-9	Lipodystrophy, familial partial, type 4 OMIM
POLD1	9175	NM_002691.4		2-27	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome OMIM
PPARG	9236	NM_138712.3		3-8	Insulin resistance, severe, digenic OMIM Lipodystrophy, familial partial, type 3 OMIM
PPP1R15B	14951	NM_032833.4		1-2	Microcephaly, short stature, and impaired glucose metabolism 2 OMIM
PTF1A	23734	NM_178161.3		1-2	Pancreatic agenesis 2 OMIM Pancreatic and cerebellar agenesis OMIM

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RFX6	21478	NM_173560.4		1-19	Mitchell-Riley syndrome OMIM
SLC19A2	10938	NM_006996.2		1-6	Thiamine-responsive megaloblastic anemia syndrome OMIM
SLC29A3	23096	NM_018344.6		1-6	Histiocytosis-lymphadenopathy plus syndrome OMIM
SLC2A2	11006	NM_001278659.2		4-10	{Diabetes mellitus, noninsulin-dependent} OMIM
SLC40A1	10909	NM_014585.5		1-8	Hemochromatosis, type 4 OMIM
STAT3	11364	NM_139276.2		2-24	Hyper-IgE recurrent infection syndrome OMIM
TFR2	11762	NM_003227.3		1-18	Hemochromatosis, type 3 OMIM
TRMT10A	28403	NM_001134665.3		2-8	Microcephaly, short stature, and impaired glucose metabolism 1 OMIM
WFS1	12762	NM_001145853.1		2-8	{Diabetes mellitus, noninsulin-dependent, association with} OMIM
YIPF5	24877	NM_030799.9		2-6	Microcephaly, epilepsy, and diabetes syndrome 2 OMIM
ZBTB20	13503	NM_015642.6		9-10	Primrose syndrome OMIM
ZFP57	18791	NM_001109809.2		1-4	Diabetes mellitus, transient neonatal 1 OMIM
ZMPSTE24	12877	NM_005857.4		1-10	Mandibuloacral dysplasia with type B lipodystrophy OMIM