Oslo universitetssykehus
Avdeling for medisinsk genetikk

Glykogenoser

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 utelatt pga. segmentale duplikasjoner.

** Transkriptets kodende ekson.

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
AGL	321	NM_000642.2		2-34	Glycogen debranching enzyme deficiency OMIM Glycogen storage disease type 3; Cori-Forbes disease; limit dextrinosis OMIM
ALDOA	414	NM_000034.3		7-14	Aldolase A deficiency OMIM Glycogen storage disease+type 12 OMIM
ENO3	3354	NM_053013.3		2-12	Enolase ? deficiency OMIM Glycogen storage disease+type 13 OMIM
G6PC	4056	NM_000151.3		1-5	Glucose-6-phosphatase deficiency OMIM Glycogen storage disease type 1a OMIM
GAA	4065	NM_000152.4		2-20	?-glucosidase deficiency OMIM Glycogen storage disease type 2; Pompe disease OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
GBE1	4180	NM_000158.3		1-16	Glycogen branching enzyme deficiency OMIM Glycogen storage disease type 4; Andersen disease; adult polyglucosan body disease OMIM
GYG1	4699	NM_004130.3		1-8	Muscle glycogenin 1 deficiency OMIM Glycogen storage disease type 15; polyglucosan body myopathy type 2 OMIM
GYS1	4706	NM_002103.4		1-16	Muscle glycogen synthase deficiency OMIM Glycogen storage disease type 0b OMIM
GYS2	4707	NM_021957.3		1-16	Hepatic glycogen synthase deficiency OMIM Glycogen storage disease type 0a OMIM
LDHA	6535	NM_005566.3		2-8	Lactate dehydrogenase A deficiency OMIM Glycogen storage disease+type 11 OMIM
NHLRC1	21576	NM_198586.2		1	Malin deficiency OMIM Progressive myoclonic epilepsy type 2B OMIM
PFKM	8877	NM_000289.5		2-23	Muscle phosphofructokinase deficiency OMIM Glycogen storage disease type 7; Tarui disease OMIM
PGAM2	8889	NM_000290.3		1-3	Muscle phosphoglycerate mutase deficiency OMIM Glycogen storage disease+type 10; DiMauro disease OMIM
PGK1	8896	NM_000291.3		1-11	Phosphoglycerate kinase deficiency OMIM
PHKA1	8925	NM_002637.3		1-32	Muscle phosphorylase kinase ?1 subunit deficiency OMIM Glycogen storage disease type 9d OMIM

Gen (HGNC symbol)	Gen (HGNC ID)	Transkript	Ekson ikke inkludert*	Ekson**	Fenotype
PHKA2	8926	NM_000292.2		1-33	Hepatic phosphorylase kinase ?2 subunit deficiency OMIM Glycogen storage disease type 9a OMIM
PHKB	8927	NM_000293.2		1-31	Phosphorylase kinase ? subunit deficiency OMIM Glycogen storage disease type 9b OMIM
PHKG2	8931	NM_000294.2		2-10	Hepatic phosphorylase kinase ?2 subunit deficiency OMIM Glycogen storage disease type 9c OMIM
PRKAG2	9386	NM_016203.3		1-16	Cardiac phosphorylase kinase deficiency OMIM
PYGL	9725	NM_002863.4		1-20	Liver glycogen phosphorylase deficiency OMIM Glycogen storage disease type 6; Hers disease OMIM
PYGM	9726	NM_005609.3		1-20	Muscle glycogen phosphorylase deficiency OMIM Glycogen storage disease type 5; McArdle disease OMIM
RBCK1	15864	NM_031229.3		1-12	HOIL1 deficiency OMIM Polyglucosan body myopathy type 1; HOIL deficiency OMIM
SLC37A4	4061	NM_001164277.1		3-11	Glucose-6-phosphate transporter deficiency OMIM Glycogen storage disease type 1b OMIM