Agilent SureSelect CD Carrier Screen Panel

Designs by experts, available to the community

Panel and design features

- Comprised of 47 genes (261.5 Kb)
- Contains coding exons with 25 bp of flanking sequence and includes select deep intronic regions
- Bears a small footprint with 70 bp samples sequenced on an Illumina NextSeq 550
- Delivers a design for equitable access to genetic identification and for multiple ethnic backgrounds, including: NW European, East Asian, Middle Eastern, African, and Ashkenazi Jewish representation
- Provides high sensitivity and specificity for single nucleotide variants (SNVs), insertions and deletions (indels), and copy number variants (CNVs)
- Uses the latest bait manufacturing improvements from Agilent, including double-stranded probes and increased uniformity
- Includes automation availability and compatibility with the Agilent Bravo NGS workstation and Agilent Magnis NGS prep system

"The benefit of the SureSelect panel-based approach is a single, unified workflow for R246 carrier testing referrals regardless of the gene of interest. This promotes efficiency through streamlining laboratory processes."

Targeted NGS assay for screening partners of carriers of known autosomal recessive disorders

Agilent Community Designs (ACDs) for next-generation sequencing (NGS) are targeted sequencing panels established in collaboration with subject matter experts in different research fields. These NGS designs are available as custom, made-to-order panels that provide you with robust and cost-effective sequencing results that focus only on your genes of interest.

NGS is now a standard part of the investigative pathway for inherited diseases. To help advance sequencing in this field, Dr. David Gokhale and Dr. Luke Stuart from the North West Genomic Laboratory Hub, Saint Mary's Hospital, Manchester, UK, have developed the Agilent SureSelect CD Carrier Screen panel.

The SureSelect CD Carrier Screen panel is designed to assay partners of carriers of a known autosomal recessive disorder where the population risk is greater than one in seventy. The pan-ethnic design facilitates equitable access to identification of gene mutations across diverse groups. Gene content has also been chosen to meet the requirements of the R246 carrier screen assay in the NHS England Genomic Test Directory. This panel ensures coverage of content relevant to the diagnostic and scientific community while maintaining the advantages of a targeted enrichment workflow.

David Gokhale, PhD,
North West Laboratory Hub,
Saint Mary's Hospital,
Manchester, UK

Luke Stuart, PhD,
North West Laboratory Hub,
Saint Mary's Hospital,
Manchester, UK

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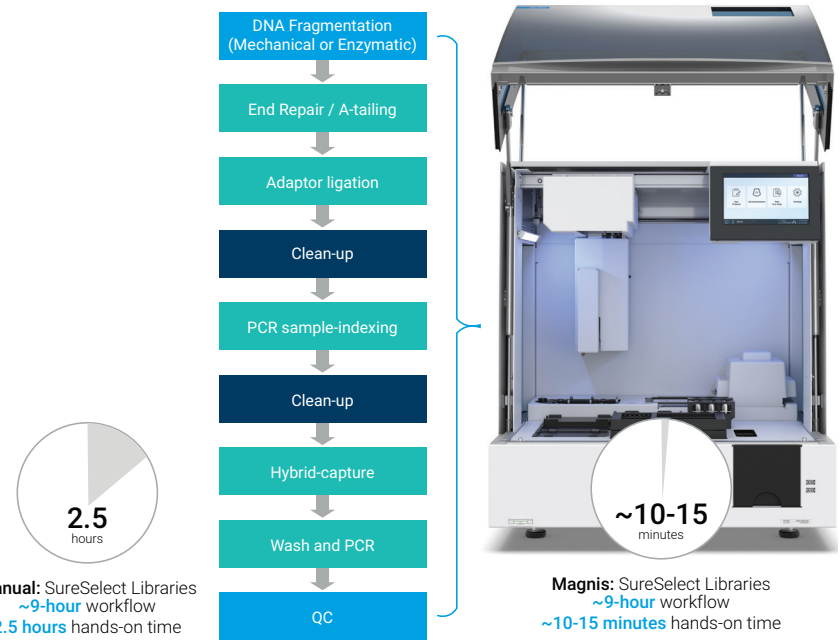
Table 1. List of genes in the SureSelect CD Carrier Screen panel.

Gene	Disease	Classified Severity
PTS	6-pyruvoyl-tetrahydropterin synthase deficiency/ hyperphenylalaninemia	Severe
ABCC8	Familial hyperinsulinism, ABCC8-related	Severe
AGA	Aspartylglucosaminuria	Severe
PKHD1	Autosomal recessive polycystic kidney disease, PKHD1-related	Severe
SACS	Autosomal recessive spastic ataxia of Charlevoix-Saguenay	Severe
CLN3	CLN3-related neuronal ceroid lipofuscinosis	Severe/profound
ASPA	Canavan disease	Profound
CPT2	Carnitine palmitoyltransferase II deficiency	Profound
PROP1	Combined pituitary hormone deficiency, PROP1-related	Moderate
OPA3	Costeff optic atrophy syndrome	Moderate
CFTR	Cystic fibrosis	Severe
ELP1/ IKBKAP	Familial dysautonomia	Severe
MEFV	Familial Mediterranean fever	Moderate
GJB2	GJB2-related DFNB1 Nonsyndromic hearing loss and deafness	Moderate
GNPTAB	GNPTAB-related disorders	Profound
GALT	Galactosemia type I	Profound
GCDH	Glutaric acidemia, GCDH-related	Moderate
G6PC	Glycogen storage disease type Ia	Severe
HBB	Hb beta chain-related hemoglobinopathy (including beta thalassemia and sickle cell disease)	Moderate
HEXA	Hexosaminidase A deficiency (Including Tay-Sachs disease)	Profound
CBS	Homocystinuria, CBS-related	Severe
MKS1	MKS1-related disorders	Profound
MMUT	Methylmalonic acidemia, MMUT-related	Profound
BCKDHA	Maple syrup urine disease type Ia	Profound
BCKDHB	Maple syrup urine disease type Ib	Profound
ACADM	Medium-chain acyl-CoA dehydrogenase deficiency	Profound
ARSA	Metachromatic leukodystrophy	Profound
NPHS1	Nephrotic syndrome, NPHS1-related	Severe/profound
PCCB	PCCB-related Propionic acidemia	Profound
PPT1	PPT1-related neuronal ceroid lipofuscinosis	Profound
SLC26A4	Pendred syndrome	Moderate
PEX6	Peroxisome biogenesis disorder type 4	Profound
PAH	Phenylalanine hydroxylase deficiency	Severe
GAA	Pompe disease	Severe
FAH	Tyrosinemia type I	Profound
ATP7B	Wilson disease	Moderate
DHCR7	Smith-Lemli-Opitz syndrome	Severe
PMM2	Congential disorder of glycosylation, type Ia	Profound
IDUA	Mucopolysaccharidosis type 1	Profound
ABCA4	Stargardt disease	Moderate
CEP290	Leber congential amaurosis 10, Joubert syndrome 5	Severe/profound
GJB6	DFNB1 nonsyndromic hearing loss and deafness (GJB6-D13S1830)del)	Moderate
CRYL1	DFNB1 nonsyndromic hearing loss and deafness (GJB6-D13S1830)del)	Moderate
GALC	Krabbe disease	Profound
IVD	Isovaleric acidaemia	Profound
BTD	Biotinidase deficiency	Profound
POLG	POLG-related disorders	Moderate/ profound

Endorsed by ACMG 2021 carrier screening guidelines.

Partner with SureSelect library preparation for optimized workflows

The Agilent SureSelect CD Carrier Screen panel can be paired with the Magnis NGS prep system and Agilent SureSelect XT HS2 kits for full walkaway automation and rapid turnaround times. SureSelect panels and baits are optimized for low input, complex samples and feature unique dual indexing, providing enhanced workflows when combined with the Bravo NGS workstation (high throughputs) or the Magnis NGS prep system (walkaway sample processing).



Manual: SureSelect Libraries
~9-hour workflow
2.5 hours hands-on time

Magnis: SureSelect Libraries
~9-hour workflow
~10-15 minutes hands-on time

Table 2. Ordering information for the SureSelect CD Carrier Screen panel. Note: All except the Magnis part numbers cover the capture probe libraries only. Library preparation and target enrichment kits must be purchased separately.

Product Name	Part Number
SureSelect CD Carrier Screen 16	5282-0089
SureSelect CD Carrier Screen 96	5282-0090
SureSelect CD Carrier Screen 96A	5282-0091
Magnis SureSelect CD Carrier Screen 32	G9694A
Magnis SureSelect CD Carrier Screen 96	G9694B

Agilent has not performed verification and validation on these panels.

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