

PRODUCT BRIEF

ASDSeq™ Research Screening Panel

RainDance Technologies ASDSeq™ Research Screening Panel*

Comprehensive panel enables scientists to identify more causative sequence mutations for autism spectrum disorder

Demonstrated Performance

The RainDance microdroplet-based platform provides unparalleled performance and a vastly simplified workflow.

- **More Complete Design Coverage:** >92% of the targeted regions
- **Greater Uniformity:** >85% of bases covered within 0.2x of the mean coverage



Overview

RainDance Technologies has collaborated with investigators at Emory University and Greenwood Genetic Center to design a comprehensive genetic screening panel for autism spectrum disorder (ASD) research. The ASDSeq™ Research Screening Panel enables researchers to simultaneously interrogate 62 genes known to affect ASD, in a single assay. Researchers can now cost-effectively apply next-generation sequencing (NGS) technology more routinely to detect known and novel mutations leading to syndromic forms of this pervasive developmental disorder.

ASD has been a highly active area of research, evidenced by the publication of more than 17,000 peer-reviewed scientific studies over the past 10 years¹. Despite the growing knowledge base in ASD, life sciences research has struggled to identify all of the causative mutations implicated in these conditions. Whole-genome sequencing is expensive and not conducive to large cohort studies; hybridization-based sequence enrichment methods and whole-exome sequencing often lack the required genomic coverage and specificity to capture all of the genes and their 5' promoter and 3' untranslated regions (UTRs). The RainDance ASDSeq panel delivers the most extensive coverage of the genes associated with this complex disorder.

The Power of RainDance

The RainDance ASDSeq Research Screening Panel provides researchers with an accurate, cost-effective, and automated genetic screening tool. The panel leverages RainDance's innovative microdroplet-based platform, which produces 1 million picoliter-size droplets per sample — each of which represents a single-molecule PCR reaction — in less than 1 hour. The power, simplicity, and precision of the RainDance technology enable researchers to solve complex biological questions with unprecedented sensitivity and specificity.

The RainDance ASDSeq™ Research Screening Panel Advantage

- **Sequence more relevant genes at the same time:** Analyze 62 genes associated with ASD in a single test with extensive coverage across coding and noncoding exons, splice junctions, and 1 kilobase (kb) of both the 5' promoter and 3' UTRs.
- **Discover more causative mutations:** Gain unprecedented single-base resolution of both common and rare genomic mutations associated with ASD.
- **Interrogate challenging regions of the genome:** Examine exons, repetitive sequences, splice sites, regulatory regions, and areas of high homology.
- **Eliminate enrichment bias:** Maintain allelic representation of heterozygous alleles with single-molecule PCR.
- **Optimize sequencer efficiency and increase sample throughput:** Reduce amount and cost of downstream NGS required with greater amplification uniformity and enrichment specificity.
- **Apply next-generation sequencing technology more routinely:** Generate accurate, consistent, and reproducible data at a fraction of the overall sample cost compared to Sanger sequencing methods.
- **Accelerate time to results and simplify workflow:** Leverage fully automated instrumentation and intuitive analysis workflow that produce more than 1 million separate PCR reactions in less than 1 hour. Decrease overall processing time to less than 8 hours compared to multiday hybridization-based sequence enrichment methods.

Comprehensive Content

The RainDance ASDSeq™ panel is the only genetic screening tool to offer greater than 92% coverage of 62 genes that contain mutations known to be associated with ASD. Coverage includes all exons for each gene plus 50 bases upstream and downstream of each exon to capture intron/exon splice junctions, as well as 1 kb of both the 5' promoter region and 3' UTRs. The genes represented in the ASDSeq Research Screening Panel include autosomal (BRAF, FOLR1, PNKP, PTPN11, SLC2A1, TCF4, and ZEB2) as well as X-linked (HPRT1 and NHS) forms of the disorder.

Autosomal Genes				
AVPR1A	EHMT1	MEF2C	PTPN11	TSC1
BDNF	FOLR1	MET	RAI1	TSC2
BRAF	FOXP1	NIPBL	RELN	UBE3A
CACNA1C	FOXP2	NRXN1	SCN1A	VPS13B
CHD7	FOXP2	NSD1	SHANK3	ZEB2
CNTNAP2	GABRB3	PFAH1B1	SLC2A1	
CREBBP	HOXA1	PNKP	SLC6A4	
DHCR7	MBD5	PTEN	TCF4	
X-Chromosome Genes				
AP1S2	DMD	L1CAM	NLGN3	PQBP1
ARX	FGD1	MECP2	NLGN4X	PTCHD1
ATRX	FMR1	MED12	OPHN1	RAB39B
CASK	HPRT1	MID1	PCDH19	SLC9A6
CDKL5	KDM5C	NHS	PHF6	SMC1A

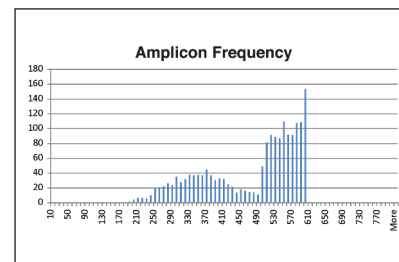


Figure 1. RainDance ASDSeq Research Screening Panel Amplicon Frequency

Table 1. Autosomal and X-chromosome genes contained in the ASDSeq Research Screening Panel.

Primer Library	Amplicons	Amplicon Bases
RainDance ASDSeq Panel	2,349	1,033,583

Table 2. RainDance ASDSeq Research Screening Panel Primer Design

Service Providers

RainDance Technologies maintains a global service provider network, including:

Expression Analysis www.expressionanalysis.com

Ambry Genetics www.ambrygen.com

Atlas Biolabs www.atlas-biolabs.de

Bioinformatics Partners

RainDance Technologies also partners with leading data analysis companies, such as:

CLCbio www.clcbio.com

Additional Research Screening Panels

RainDance Technologies also offers comprehensive screening panels for x-chromosome and human leukocyte antigen (HLA) and major histocompatibility complex (MHC) research. The RainDance Xeq™ panel simultaneously interrogates more than 800 genes on the X chromosome and the HLASeq™ panel targets the entire HLA super locus (3.8 Mb region).

References

1. <http://www.ncbi.nlm.nih.gov/pubmed>

**The RainDance ASDSeq™, XSeq™, and HLASeq™ Research Screening Panels are for research use only. Not for diagnostic purposes.*

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