

## Ashkenazi Jewish Panel

# SPOT ON

## INFINITI™

*The Automated Multiplexing MDx Solution*



### Product Design

- ▶ The INFINITI™ System Assay for Ashkenazi Jewish Panel is designed for the detection of genetic disorders in the Ashkenazi Jewish population.
- ▶ The INFINITI System Assay for Ashkenazi Jewish Panel utilizes the Ashkenazi Jewish Panel Intellipac™, Ashkenazi Jewish Panel Amp Mix and Ashkenazi Jewish Panel BioFilmChip™ Microarray.
- ▶ The INFINITI System Assay for Ashkenazi Jewish Panel is automated by the 510(k) cleared INFINITI Analyzer.
- ▶ Clinical validation is currently in progress.

### Benefits

	VERSATILITY	◆	Multiplexed determination of 31 genetic variants for genetic disorders affecting Ashkenazi Jewish individuals
	EFFICIENCY	◆	Rapid turnaround time enhances workflow efficiency
	AGILITY	◆	<i>Load N Go</i> automation with the INFINITI Analyzer
	INTEGRITY	◆	Replicate determinations on a single BioFilmChip Microarray ensure quality results

### Genetic Variants

Canavan:	Y231X, E285A, A305E, and 433 (-2) A/G
Gaucher:	N370S, V394L, D409H, L444P, R496H, IVS2 (+1)G/A, 84 insG/GG, and 1263-1317del55
Familial Dysautonomia:	R696P, IVS20 (+6) T/C
Nieman Pick:	L302P, (-1) del p330FS, R496L, and R608 (-3)
Fanconi Anemia:	322delG, IVS4(+4)A/T
Bloom Syndrome:	2281delATCTGAinsTAGATTC
Tay Sachs:	R247W, R249W, G269S, 1278 insTATC, IVS9(+1)G/A, IVS12(+1)G/C, and 7.6kb del
Mucopolipidosis IV:	IVS3 (-2)A/G, del6.4kb

### Sample Type and Volume

0.2 – 2.0 ml of peripheral whole blood in EDTA (purple-top) tube  
50 ng DNA / reaction

### Product Information

Product No.	Product Name	Description	Pack Size
01 107	INFINITI Ashkenazi Jewish Panel BioFilmChip	12 BioFilmChips / Magazine	4 Magazines / pack
01 207	INFINITI Ashkenazi Jewish Panel Intellipac	24 tests / IntelliPac	2 Intellipac / pack
01 307	INFINITI Ashkenazi Jewish Panel Amp Mix	250 µl / vial	4 vials / pack

Please contact AutoGenomics to obtain product information and for product status updates.

SM-40021 / 0408

1-866-STAT-MDx

www.autogenomics.com





### Clinical Relevance

- ▶ The Ashkenazi Jewish Panel is designed to detect genetic mutations in several genes which are responsible for single-gene disorders and disease predispositions that are higher in the Ashkenazi Jewish population, but not exclusive to this group.
- ▶ These diseases are autosomal recessive inherited traits, and the majority of them are severe.
- ▶ The presence of some of these mutations determine whether or not an individual is a carrier of a defective gene, with the risk of transmitting a defective gene to his or her descendants.
- ▶ Since all of these disorders are autosomal recessive, if both parents are carriers, they are at risk of having an affected child.

### Clinical Utility

- ▶ Screening is recommended for Ashkenazi Jews and their partners who are expecting a child or contemplating pregnancy.
- ▶ If one partner is Ashkenazi Jewish and the other is not, screening of the couple is recommended beginning with the Ashkenazi Jew.<sup>1</sup>

### Reference Data<sup>2</sup> :

Disease (Symbol)	Mutations Screened	Ashkenazi Disease Incidence	Ashkenazi Carrier Frequency
Bloom ( <i>BLM</i> )	2281del6/ins7	1/40,000	1/100
Canavan ( <i>ASPA</i> )	Y231X, E285A, A305E, 433(-2)A>G	1/10,000	1/50
Familial Dysautonomia ( <i>IKBKAP</i> )	R696P IVS20(+6)T>C	1/3,600	1/32
Fanconi Anemia Group C ( <i>FANCC</i> )	322delG IVS4(+4)A>T	1/32,000	1/89
Gaucher ( <i>GBA</i> )	84G>GG, N370S IVS2(+1)G>A, V394L, del55bp, D409H, L444P, R496H	1/900	1/15
Mucopolipidosis IV ( <i>MCOLN1</i> )	del6.4kb IVS3(-2)A>G	1/63,000	1/127
Niemann-Pick Type-A ( <i>SMPD1</i> )	L302P, R496L, 1bp del fsP330, R608del	1/32,000	1/90
Tay-Sachs ( <i>HEXA</i> )	del7.6kb, R249W, G269S, IVS9(+1)G>A 1278insTATC, IVS12(+1)G>C, R247W	1/3,000	1/30

### References

1. <http://www.questdiagnostics.com/hcp/topics/ashkenazi/ashkenazi.html>
2. ARUP Laboratories Test Directory, Ashkenazi Jewish Panel <http://www.aruplab.com/guides/ug/tests/0051415.jsp>