savyonDIAGNOSTICS

available Q1 2013

NanoCHIP®

NanoCHIP All In One Kit

Cat #800014

The "All In One" kit is used to detect and identify a panel of the most common 27 genetic diseases (92 mutations all together) in the Jewish Israeli population. The assay is preformed on the NC400 Automated Microarray platform using the NanoChip microarray cartridge.

Name Of Disease	Full disease and genes name	Mutations list	Number of mutations
Ashkenazi Jews			
CF17	Cystic Fibrosis (CFTR)	F508 • S549R • G542X • G85E • W1282X • W1089X • N1303K • 405+1 G>A • 3849+10KB C>T • Y1092X • 1717-1 G>A • 3121-1G>A • D1152H • I1234V • Del2,3 (21Kb) • Q359K/360K • 2751+1insT	18
AJP	Alpha 1 Antitripsin (SERPINA1) • Bloom Syndrome (BLM) • Fanconi Anemia C (FANCC) • Familial Dysautonomia (IKBKAP) • Glycogen Storage Type 1a (G6PC) • Mucolipidosis IV (MCOLN1) • Maple Syrup Disease (BCKDHB) • Niemann Pick Type A,B (SMPD1) • Usher Syndrome (PCDH15) • Canavan (ASPA) • Nemaline (NEB) • Jubert (TMEM216)	PiZ PiS • 6bp del/7,bp Ins • IVS4+4A>T • R696P G>C • 2507+6 ,T>C • R83C • IVS3-2A>G • Del(EX1-EX7) • R183P • fsP330 • L302P • R496L • R608 • R245X • 854A>C • 693C>A • R2478-D2512del • c.35G>T (R12L)	19
GSD1a	Glycogen Storage Type 1a (G6PC)	Q347X (c.1118C>T or c.1039C>T)	1
Bloom	Bloom Syndrome (BLM)	2407-2408dupT	1
MSUD	Maple syrup urine disease (BCKDHB)	G278S EXON 7 • E372X EXON 10	2
Connexin	Hereditary Hearing Loss (GJB2)	35del • 167delIT • Cx30 del	3
Tay Sachs	Tay Sachs (HEXA)	1278Ins • IVS12+1 • G269S • R170Q • IVS5-2A>G • F304	7
Gaucher	Gaucher Type 1 (GBA)	84GG • N370S • IVS2+1 • V3942 • L444P • R496H • RecTL	7
DLD	Dihydrolipoyl dehydrogenase deficiency (DLD or LAD)	G229C • Y35X	2
Galactosemia	Galactosemia (GALT)	5.5 kb complex deletion K285N	2
USH3A	Usher Syndrome type III A (CLRN1)	N48K (c.144T>G)	1
Thyrosinemia	Thyrosinemia (FAH)	P261L	1
North African Jews			
AT	Ataxia telangiectasia (ATM)	R35X (c.103C>T)	1
GSD 3	Glycogen storage disease IIIa (AGL)	4455delT	1
Fanconi A	Fanconi anemia type A (FANCA)	2173/3insG • 4275delT	2
LGMD2b	Limb-girdle muscular dystrophy type 2b (DYSF)	1624delG (*available also at Jews from daghastan)	1*
MLC1	Megalencephalic leukoencephalopathy with subcortical cysts 1 (MLC1)	G59E (c.176G>A)	1
CTX	Cerebrotendinous Xanthomatosis (CYP27A1)	T339M (c.1016C>T) • 1253delT • IVS4-1G>A	3
PCCA	Progressive cerebello-cerebral atrophy (SEPSECS)	A239T (c.715G>A)	1
TMC1	Hearings loss (TMC1)	R604X (c.1810X>T) • S647P (c.1939T>C • R389X (c.1165C>T) • W404R (c.1210T>C)	4
Yamane Jews			
MLD	Metachromatic leukodystrophy (ARSA)	P377L (c.2119C>T)	1
PKU	Phenylketonuria (PAH)	DEL 6296 BP	1
RP26	Retinitis Pigmentosa (CERKL)	(c.238+1G>A)	1
Iraqi Jews			
3MGA (Kostaf)	Optic Atrophy Syndrome 3 (OPA3)	IVS-1G>C	1
PCCA	Progressive cerebello-cerebral atrophy (SEPSECS)	p.Y334C (c.1001A>G)	1
Connexin 26	Non syndromic Hereditary Hearing Loss (GJB2)	L90P (c.269T>C) In addition to the common 3 mutations	1
Tay Sachs	Tay Sachs disease (HEXA)	G250V (c.G749T) • L451V (c.C1351G) • R393X (c.1177C>T)	3
Usher syndrome	Usher syndrome IIa (USH2A)	c.236_239dupGTAC	1
Bukhara-Uzbekistan			
Connexin 26	Non syndromic Hereditary Hearing Loss (GJB2)	51del12insA	1
Persian, Karaite Jews / Beduin, Palestinian (Moslems)			
HIBM	Hereditary Inclusion body myopathy (GNE)	M712T (c.2186T>C)	1
Non Jews (Christian)			
Tay Sachs	Tay Sachs disease (HEXA)	IVS9+1G>A	1
Jews from daghastan - Caucasus			
ICCA	Infantile cerebral cerebellar atrophy (MED 17)	L371P	1
LGMD2b	Limb-girdle muscular dystrophy type 2b (DYSF)	1624delG (*available also at Jews from North Africa)	1*
Total			92

THE STANDARDS INSTITUTION OF ISRAEL

www.nanochip400.com

It's as simple as **1,2,3...**

PCR mix1
PCR mix2
PCR mix3

Master Mix

PCR plate

PCR

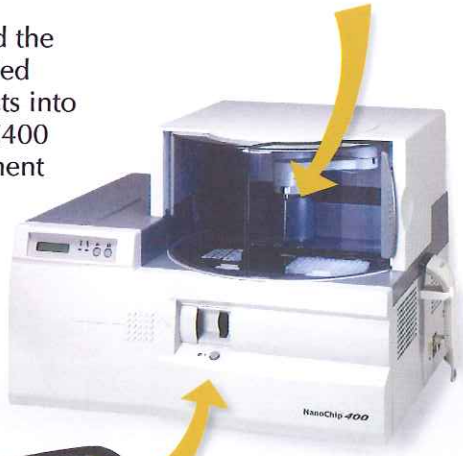
1. Prepare 3 PCR mixes for each sample

Reagent Pack

2. Seal the PCR plate with the MicroPlate seals and load into the thermal cycler

3. Run the PCR program (1.5 hours)

4. Load the amplified products into the NC400 instrument



NC400 Instrument

NanoChip® Cartridge

5. Scan and load reagent cassettes and buffer bottles

6. Scan and insert cartridge. Start the run (9.5 hours walk-away automation for 24 AIO samples)

- Full genotype of 27 different diseases, 92 mutations - all from single DNA sample in one test!
- Fully automated NanoChip® Microarray platform.
- Short 1.5 hour Multiplex PCR in 3 vials.
- Up to 24 samples per run in the same day (9.5 hours per run).
- User Friendly operation and minimal hands on time.
- LIS connectivity.
- Additional CE-IVD assays are available for various applications such as: Genetic Screening, PG'x, Infectious Diseases and more.
- CE-IVD approval in process.

7. Analyze data and export it to LIS

Position	Sample ID	Data Set	ML4-del	ML4-IVS3	NPA-L302P	NPA-R496L
1	Sample 1	AIO	HET	-	-	-
2	Sample 2	AIO	-	-	-	-
3	Sample 3	AIO	-	HET	-	-
4	Sample 4	AIO	-	-	HOM	-

Partial All In One assay results demonstrated

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