



Wider view on mutation spectrum

Ashkenazi Jewish Diseases Analysis Kit

– all you need in one box –

Quick facts

- 78 genetic variations from 23 genes detected simultaneously in a single run
- Variations cover Ashkenazi and Sephardic Jewish populations
- Developed with the leading scientists in the Ashkenazi Jewish Diseases research community backed by the latest scientific and medical studies

Easy to use

- Everything you need is in one box
- Analysis software comes with integrated guidance for mutation detection
- Barcoded arrays to ensure precise tracking
- Consultations from day-to-day users

Ashkenazi Jewish Diseases

- Genorama reliable Ashkenazi Jewish Diseases Analysis Kit is based on flexible and cost-effective Arrayed Primer EXTension (APEX) genotyping technology
- Enables to determine 78 disease related genetic variations in 23 genes - HEX A, BLM, ASPA, SMPD1, IKBKAP, DYT1, MCOLN1, FANCC, F11, G6PC, BCKDHB, GJB2, MEFV, GDE, GBA, CFTR, SERPA, NEB, PCDH15, ABCC8, LDLR, DLD and TMEM216
- Cystic Fibrosis, Tay-Sachs Disease, Bloom Syndrome, Canavan Disease, Niemann-Pick A, Familial Dysautonomia, Torsion Dystonia, Mucopolidosis type IV, Fanconi Anemia, Gaucher Disease, Factor XI Deficiency, Glycogen Storage Disease Type 1a, Maple Syrup Urine Disease, Non-Syndromic Sensorineural Hearing Loss, Familial Mediterranean Fever, Alpha 1-anti-trypsin Deficiency, NemaLine Myopathy, Usher Syndrome Type 1F, Familial Hyperinsulinemia, Familial Hypercholesterolemia, Lipoamide Dehydrogenase Deficiency, Glycogen Storage Disease type III and Joubert Syndrome 2
- Genorama Ashkenazi Jewish Diseases test is suitable for both carrier screening and confirming the diagnosis

Ashkenazi Jewish Diseases Analysis Kit

– all you need in one box –

Reliable and up to date

- >99.9% accuracy
- List of relevant publications is available on Genorama website
- Intensively used throughout years and hundreds of screened samples
- Based on established and validated APEX technology
- Genorama is keeping a close eye on new discoveries in the field

Genorama Ashkenazi Jewish Diseases Analysis KIT includes all consumables for successful analysis process and provides all necessary reagents, PCR primers and barcoded arrays for 25 tests

- PCR primers and purification Kit
- APEX Template Preparation Kit
- APEX Reaction Mixture Kit
- Ashkenazi Jewish diseases microarrays

APEX technology

Arrayed Primer EXTension (APEX) genotyping is based upon an array of oligonucleotides, immobilized on glass surface via their 5' end. Patient's DNA is amplified by PCR, digested enzymatically and annealed to the immobilized primers, which promote sites for template-dependent DNA polymerase extension reactions using four unique fluorescently labelled dideoxy nucleotides. The mutation is detected by the change in primer sites colour code using the previously installed Genorama genotyping platform.

Ordering information

Cat #	Product name
G60109	Ashkenazi Jewish Diseases Analysis KIT (25 reactions)

For further information please contact Genorama representatives

Genetic variations in test

HEX A 1278insTATC; 1421+1G>C; 805 G>A; IVS9+1G>A; 745 C>T; 739 C>T; IVS7+1G>A

BLM Δ7.6kb; 2281del6/ins7 (from start codon 2206del6/ins7)

ASPA 433-2A>G; 693 C>A, polymorphism C>T; 854A>C; 914C>A

SMPD1 905T>C; 330delC; 1487G>T; ΔR608 (N-P B)

IKBKAP 2087G>C; 2204+6T>C

DYT1 ΔE302/303 (c301 ensemble); ΔF323-Y328 (18 bp del)

MCOLN1 IVS3-2A>G; 511del6944

FANCC 37C>T; 322ΔG; 711+4A>T; 1897 C>T also in AJ's =1689C>T (1897C>T), =1642C>T Ensembles

F11 576G>T; 1074A>C; IVS14+1G>A

G6PC 247C>T

BCKDHB 548 G>C; 832G>A; 1114G>T

GJB2 35delG; 167delT

MEFV 2080A>G

GDE or Amylo-1,6-glucosidase Δ4455T

GBA 84 insG; IVS2+1G>A; 1035insG; 1376A>G (also1226A>G); 1297G>T (also 1307G>T); 1448T>C; 1604G>A

CFTR 386G>A; 482G>A/C/T; 621+1G>T; 711+1G>T; 1132C>T; 1172G>A/C/T; 1496C>A; DeltaI507; DeltaF508; 1717-1G>A, G>T; 1756G>T; 1784G>A; 1789C>T/G; 1811G>C/A; 1898+1G>A/T/C; 2184delA; 2183AA>G; 2789+5G>A; 3120+1G>A; 3616C>T; 3659delC; '3849+10kbC>T; 3978G>A/T; 4041C>G

SERPA c268 GAG->AAG

NEB delIVS54-2025 to IVS55+372

PCDH15 733C>T

ABCC8 delTTC; 3922-9g>a

LDLR 652delGGT

DLD 105insA; c770 G>T

TMEM216 c.35G>T or c.218G>T