ARUPConsult

Ashkenazi Jewish Genetic Diseases Carrier Screening Algorithm

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INDICATION FOR TESTING
Ashkenazi Jewish ancestry

Comprehensive Ashkenazi Jewish carrier testing desired

Yes

ORDER
Ashkenazi Jewish Diseases, 16 Genes (*BLM, ASPA, IKBKAP, FANCC, GBA, MCOLN1, SMPD1, HEXA, ABCC8, G6PC, TMEM216, BCKDHB, PCDH15, CLRN1, DLD,* and *NEB*)
AND
Cystic Fibrosis (*CFTR*) 165 Pathogenic Variants

Targeted molecular analysis for 16 disorders common in individuals of Ashkenazi Jewish descent

- Bloom syndrome
- Canavan disease
- Familial dysautonomia
- Fanconi anemia group C
- Gaucher disease
- Mucolipidosis type IV
- Niemann-Pick disease type A
- Tay-Sachs disease
- *ABCC8*-related hyperinsulinism
- Glycogen storage disease type 1A
- Joubert syndrome type 2
- Maple syrup urine disease type 1B
- Usher syndrome type 1F
- Usher syndrome type 3
- Lipoamide dehydrogenase deficiency
- *NEB*-related nemaline myopathy

No

INDICATIONS FOR TESTING

- French-Canadian ancestry
- Louisiana Cajun ancestry
- Positive family history for Tay-Sachs disease
- Partner is a confirmed carrier of Tay-Sachs disease

Screening for Tay-Sachs disease only

ORDER
Hexosaminidase A Percent and Total Hexosaminidase, Plasma or Serum (for males, nonpregnant women, and women not taking oral contraceptives)
OR
Hexosaminidase A Percent and Total Hexosaminidase in Leukocytes (for pregnant women or women taking oral contraceptives)

Hexosaminidase A enzyme quantitation is inconclusive or indicates carrier

ORDER
Tay-Sachs Disease (*HEXA*), 7 Variants

Variant detected (carrier)

Test partner
ORDER
Hexosaminidase A Percent and Total Hexosaminidase, Plasma or Serum (for males, nonpregnant women, and women not taking oral contraceptives)
OR
Hexosaminidase A Percent and Total Hexosaminidase in Leukocytes (for pregnant women or women taking oral contraceptives)

No variant detected

CONSIDER

- *HEXA* gene sequencing
- Testing partner

Pseudodeficiency variant detected (noncarrier)

Hexosaminidase A enzyme quantitation indicates noncarrier

Hexosaminidase A and total enzyme quantitation indicates possible Sandhoff carrier

Test partner for Sandhoff carrier status

INDICATIONS FOR TESTING

- Family history of a Jewish genetic disorder other than Tay-Sachs
- Partner is a carrier of a Jewish genetic disorder other than Tay-Sachs

Offer DNA testing to assess carrier status for the familial disorder or the disorder for which the partner is a carrier

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