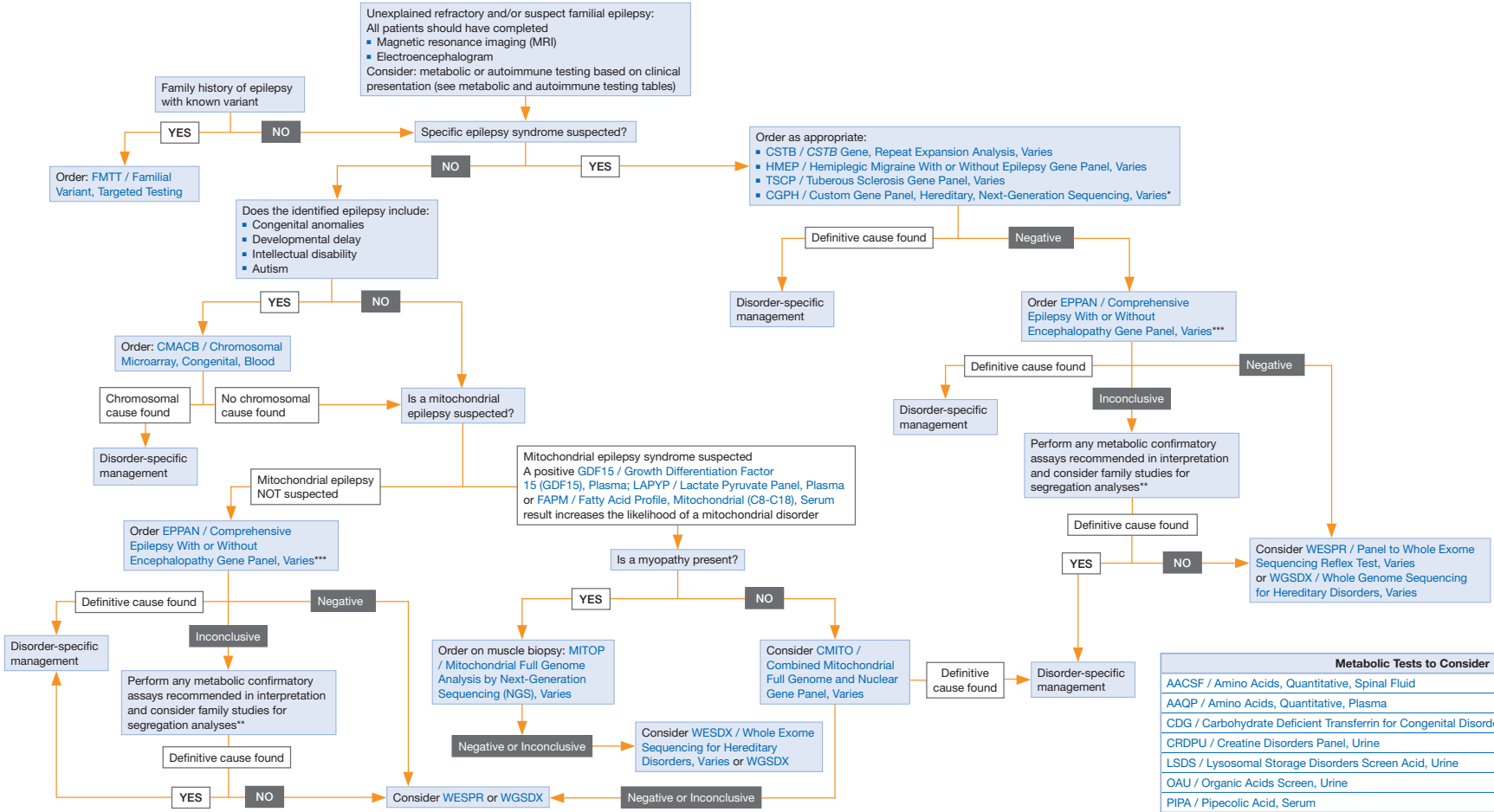
MAYO CLINIC LABORATORIES

Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm



Clickable PDF
Click on blue tests to view test catalog information

Metabolic Tests to Consider
AACSF / Amino Acids, Quantitative, Spinal Fluid
AAQP / Amino Acids, Quantitative, Plasma
CDG / Carbohydrate Deficient Transferrin for Congenital Disorders of Glycosylation, Serum
CRDPU / Creatine Disorders Panel, Urine
LSDS / Lysosomal Storage Disorders Screen Acid, Urine
OAU / Organic Acids Screen, Urine
PIPA / Pipecolic Acid, Serum
PIPU / Pipecolic Acid, Urine, if newborn
PLSD / Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot, if <18 years of age
POXP / Fatty Acid Profile, Peroxisomal (C22-C26), Plasma
PUPYP / Purines and Pyrimidines Panel, Plasma
PYRC / Pyruvate, Spinal Fluid
Autoimmune Evaluations to Consider
EPC2 / Epilepsy, Autoimmune/Paraneoplastic Evaluation, Spinal Fluid
EPS2 / Epilepsy, Autoimmune/Paraneoplastic Evaluation, Serum

*CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies can be used to modify any epilepsy panel or test any single gene included on any epilepsy panel.

**Segregation studies can be performed to determine if a variant segregates with the condition in a family and/or occurred *de novo*, which may clarify the significance of a variant. For more information, contact the Laboratory Genetic Counselors at 800-533-1710.

***WESDX / Whole Exome Sequencing for Hereditary Disorders, Varies or WGSDX / Whole Genome Sequencing for Hereditary Disorders, Varies could be considered as an alternative first tier test.