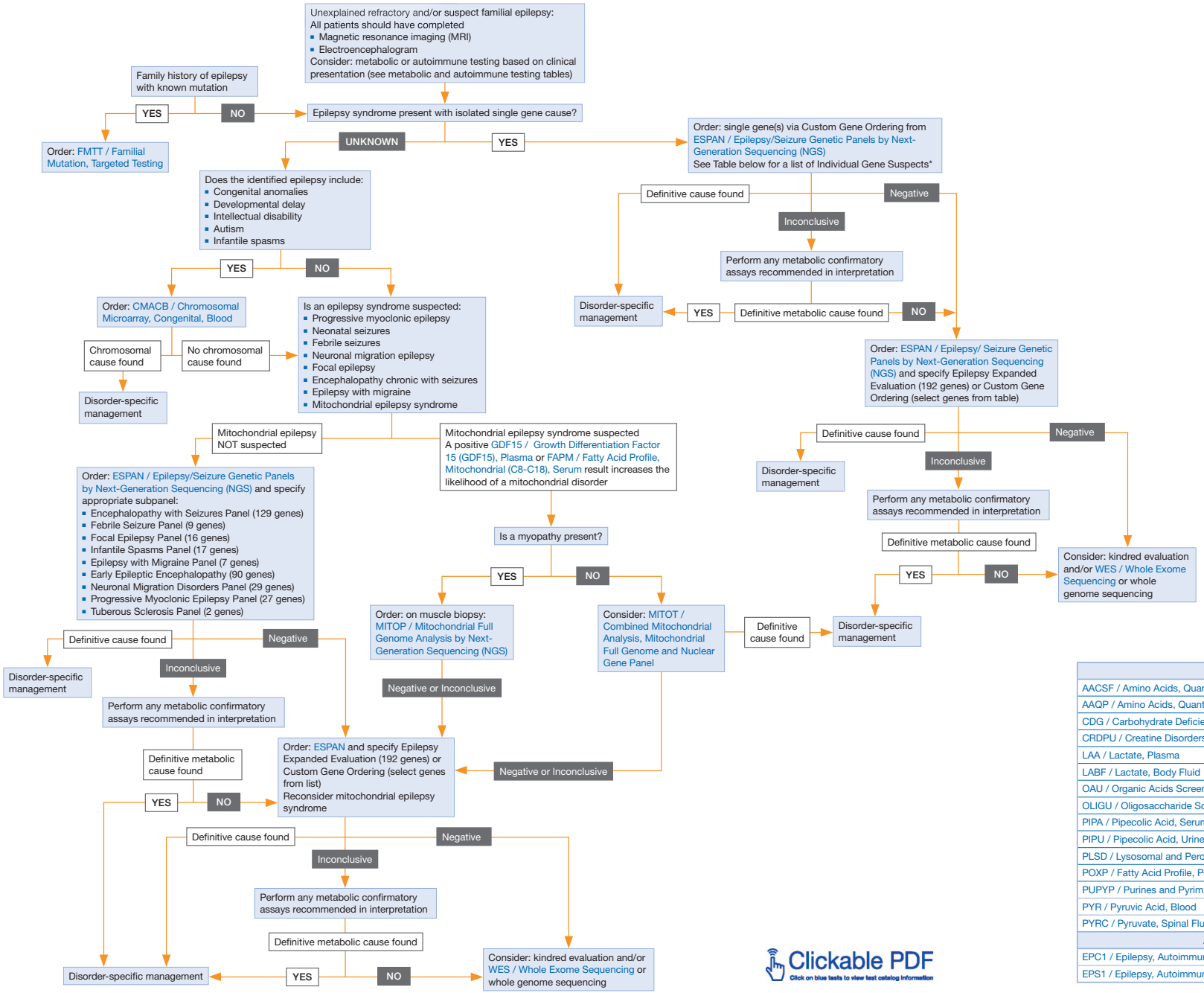
MAYO CLINIC LABORATORIES

Epilepsy: Unexplained Refractory and/or Familial Testing Algorithm



*Individual Gene Suspects		
Type	**Gene**	**OMIM**
Ceroid lipofuscinosis, neuronal, 2	*TPP1*	204500
Ceroid lipofuscinosis, neuronal, 3	*CLN3*	204200
Dravet syndrome, severe myoclonic epilepsy of infancy	*SCN1A*	607208
Encephalopathy epileptic, early infantile	*PCDH19*	300088
Encephalopathy epileptic, early infantile; seizures, benign neonatal	*KCNQ2*	613720; 121200
GLUT1-deficiency syndrome	*SLC2A1*	606777
Infantile spasms	*ARX*	308350
Myoclonic epilepsy progressive	*CSTB*	254800
Myoclonus-nephropathy	*SCARB2*	254900
Pyridoxine-dependent epilepsy	*ALDH7A1*	266100
Rett syndrome (order as MECPZ / MECP2 Gene, Full Gene Analysis)	*MECP2*	312750
Seizures, benign neonatal	*KCNQ3*	121201
Temporal epilepsy familial	*LGI1*	600512

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
LAA / Lactate, Plasma
LABF / Lactate, Body Fluid
OAU / Organic Acids Screen, Urine
OLIGU / Oligosaccharide 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
PYR / Pyruvic Acid, Blood
PYRC / Pyruvate, Spinal Fluid
Autoimmune Evaluations to Consider
EPC1 / Epilepsy, Autoimmune Evaluation, Spinal Fluid
EPS1 / Epilepsy, Autoimmune Evaluation, Serum

Clickable PDF
Click on blue tests to view test catalog information

Custom Gene Ordering is available at www.MayoClinicLabs.com