Supplemental Table e-1. List of all testing performed.

Adrenocorticotropic Hormone (ACTH)

Aldolase

Alpha Fetoprotein

Amino Acids (plasma)

Ammonia

Biotinidase

Bone Marrow Biopsy

Carbohydrate Deficient Transferrin/Glycoprotein

Cardiolipin Ab

Carnitine Studies

Complete blood count

Ceruloplasmin

Chemistry Panel

Chemistry - Minerals

Chromosomal Analysis (Banding or FISH)

Complement Studies

Congenital Infections Tests

Cortisol

Creatine Kinase

CRP (not for CV risk)

CSF routine analysis

CSF amino acids

CSF immunoglobulins

CSF lactate

CSF neurotransmitters and metabolites

Drug screen

Fat, fecal

Fibroblast culture

Folate (Vitamin B9)

GI disorders panel

Glutamic Acid Decarboxylase Ab

Growth Studies panel

Hepatic Function

HLA Antibody Screen

Homocysteine

Homovanillic acid and Vanillylmandelic acid

Immunoglobulins (serum)

Urine organic acids

Infectious Meningoencephalitis Tests

Insulin Interferon Alpha

Ion Channel Antibodies

Ketone Bodies

Lactate

LDH

Lead

Leukocyte Lysosomal Enzymes

Lipid Studies

Lymphocyte subset panel 7 - congenital immunodeficiencies

Methylmalonic Acid

Microarray (CGH or SNP)

Mucopolysaccharide Screen

MPO

Neutrophil Receptor Panel

Nuclear Ab

Oligosaccharide Screen

Organic Acids (urine or plasma)

Parathyroid Hormone

Peroxisomal Panel

Pipecolic Acid

PKU Cofactor Screen

Pyruvate

Reducing Substances (urine or feces)

Rheumatoid Factor

Sedimentation Rate

Selenium

Sialic Acid (urine or lipid-associated in plasma)

Telomeres

Thyroid Studies

Uric Acid

Urinalysis

Urinary Purine Panel

Urinary Pyrimidine Panel

Vitamin A

Vitamin B12

Vitamin E

3-Methylglutaconic aciduria

VLCFA

Alexander Disease (GFAP)

Alpha-1-Antitrypsin Deficiency

Angelman Syndrome

Arginine:glycine aminidinotransferase deficiency (AGAT)

ARX disorders

Autoimmune limbic encephalitis (N-methyl-D-aspartate receptor ab)

Batten Disease - Infantile Neuronal Ceroid Lipofuscinosis (CLN1)

Batten Disease - Late-Infantile Neuronal Ceroid Lipofuscinosis (CLN2)

Batten Disease - Juvenile Batten Disease (CLN3)

CGH1 diseases

CMT Disease

Cystic Fibrosis (Sweat Test or mutation panel)

FOXG1 Syndrome

Fragile X Syndrome

Fucosudosis

GM1 Gangliosidosis

Hypomyelinating Leukodystrophy 8 (POLR3B)

Infantile Neuroaxonal Dystrophy (PLA2G6)

Krabbe Disease (GALC)

Leigh Disease

MeCP2 Diseases (including Rett Syndrome)

Metachromatic Leukodystrophy (ARSA or Sulfatide Extraction)

Mitochondrial Panel/MTDNA evaluation

Multiple Sclerosis Panel

Myositis Antibody Panel

Neuromyelitis Optica (Aquaporin antibodies)

Pelizaeus Merzbacher Disease (PLP)

Pelizaeus Merzbacher-Like Disease (PMLD, gene GJC2)

Periodic Paralysis - (Hyperkalemic (SCN4A), Hypokalemic (SCN4A E12, CACNA

1S),Normokalemic (SCN4A E13))

POLG Testing

Pyruvate Carboxylase or Pyruvate Dehydrogenase

Sandhoff Disease Gene Sequencing (HEXB)

SCN1A disorders

Systemic Lupus erythematosus (ribosomal p antibody)

Smith-Lemli-Opitz Syndrome (7 dehydrocholesterol)

Sotos Syndrome (NDS1)

Spinal Muscular Atrophy (SMN)

S-Sulfocysteine disorders

Treacher Collins-Franceschetti syndrome 1

Tyrosine Hydroxylase Deficiency

Vanishing White Matter Disease (EIF2B)

X-linked Infantile Spasm Syndrome (CDKL5)