VIRTUAL MEDICAL PRACTICE, LLC
expanding genetic horizons……

Possible Symptoms of Mitochondrial Disease

BRAIN

- Developmental delays
- Dementia
- Neuro-psychiatric disturbances
- Migraines
- Autistic Features
- Mental retardation
- Seizures
- Atypical cerebral palsy
- Strokes

NERVES

- Absent reflexes
- Weakness (may be intermittent)
- Fainting
- Dysautonomia - temperature instability & other dysautonomic problems
- Neuropathic pain

MUSCLES

- Weakness
- Cramping
- Gastrointestinal problems
- Dysmotility
- Irritable bowel syndrome
- Hypotonia
- Muscle pain
- Gastroesophogeal reflux
- Diarrhea or constipation
- Pseudo-obstruction

KIDNEYS

- Renal tubular acidosis or wasting

HEART

- Cardiomyopathy
- Cardiac conduction defects (heart blocks)

LIVER

- Liver failure
- Hypoglycemia (low blood sugar)

EARS & EYES

- Visual loss and blindness
- Ptosis
- Ophthalmoplegia
- Optic atrophy
- Hearing loss and deafness
- Acquired strabismus
- Retinitis pigmentosa

PANCREAS & OTHER GLANDS

- Diabetes and exocrine pancreatic failure (inability to make digestive enzymes)
- Parathyroid failure (low calcium)

SYSTEMIC

- Failure to gain weight
- Fatigue
- Unexplained vomiting
- Short stature
- Respiratory problems

Mail: 5579 Chamblee Dunwoody Rd, Suite 110, Atlanta, GA 30338
Clinic: 3400 Old Milton Pkwy, Bldg A, Ste 500, Alpharetta, GA 30005
Voice: 404.720.0820 Fax: 404.601.9931 www.virtualmdpractice.com

VIRTUALMEDICALPRACTICE, LLC
expanding genetic horizons……

Genetic Testing for Autistic Spectrum Disorder (ASD) Patients

Although *some* of the first tier tests can be obtained without a subspecialist's input, interpretation of the data may be difficult without the involvement of a genetics specialist. Decisions regarding whether a specific patient requires a more in-depth investigation as shown in the second tier tests for mitochondrial or other rare metabolic or genetic diseases should be undertaken by a mitochondrial expert and/or a biochemical geneticist. Such a decision should be based on a number of factors, including screening results, laboratory testing, family history, physical findings, and clinical features. In general, the genetics workup and ongoing management of an ASD patient (should a genetics diagnosis be made) is best completed by someone trained in genetics with mitochondrial and metabolic disease experience and expertise.

TIER 1 - basic work-up recommended for all patients
▪ Chromosome microarray studies ▪ Fragile X ▪ Complete metabolic panel, CBC, CPK ▪ Ammonia level ▪ Lactate and pyruvate levels ▪ Carnitine, plasma total and free ▪ Coenzyme Q10 level ▪ Plasma and urine amino acids ▪ Urine organic acids ▪ Plasma acylcarnitines ▪ Thyroid function tests
TIER 2 - depends on clinical features and results of Tier 1 testing
▪ Mitochondrial enzyme and/or DNA testing ▪ Rett syndrome DNA testing ▪ Atypical Rett (CDKL5 gene testing) ▪ PTEN mutational analysis ▪ NLGN3, NLGN 4x, SHANK3, SNRPN gene testing ▪ Lysosomal enzyme testing ▪ Peroxisome disease testing (VLCFAs) ▪ CSF studies for lactate and pyruvate, amino acids and neurotransmitters ▪ Brain MRI

Mail: 5579 Chamblee Dunwoody Rd, Suite 110, Atlanta, GA 30338
Clinic: 3400 Old Milton Pkwy, Bldg A, Ste 500, Alpharetta, GA 30005
Voice: 404.720.0820 Fax: 404.601.9931 www.virtualmdpractice.com