POTOCKI-LUPSKI SYNDROME (PTLS)
GUIDELINES FOR YOUR HEALTH CARE PROVIDERS

The Potocki-Lupski syndrome (PTLS) is a chromosomal duplication syndrome (duplication 17p11.2) with an incidence of at least 1 in 20,000. Most individuals with PTLS have only mildly dysmorphic features, thus a chromosomal abnormality is usually not suspected clinically. Most children diagnosed with PTLS present with developmental delay, speech and language delay, and autistic-like behavior. Other common features of PTLS (seen in 70%-100% of patients) include failure to thrive, oral-pharyngeal dysphasia, hypotonia, abnormalities on electroencephalogram, hypermetropia on ophthalmic examination, and sleep disordered breathing. Structural cardiac anomalies are observed in 50% of patients and include left ventricular outflow tract anomalies such as bicuspid aortic valve, dilated aortic root, and hypoplastic left heart.

The following are general healthcare guidelines for individuals with PTLS based on my experience as a physician and medical geneticist. These guidelines are intended to assist physicians and health care providers in determining the best management for individuals with PTLS and can never replace good clinical judgment and practice. Most of the studies below should be followed and re-assessed as the child grows and develops. Any abnormalities should be addressed and managed medically or surgically as indicated.

- Feeding evaluation; assessment for failure to thrive if applicable
- Developmental assessment
- Autism spectrum disorder assessment (ADI and ADOS)
- Speech and language assessment
- Ophthalmological examination
- Otolaryngological examination (ENT)
- Audiological examination
- Echocardiogram and evaluation of the aortic root
- Electrocardiogram
- Renal ultrasound
- Sleep study (to assess for sleep apnea)

On-going therapies should include:

- Speech and language therapy
  With consideration for augmentative communication.
- Physical therapy
  With consideration for adaptive physical education
- Occupational therapy

or contact:

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