POTOCKI-LUPSKI PTLS SYNDROME

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MEDICAL RECOMMENDATIONS

Physician	Evaluation to be done	Comments

Infants and Children
 	
Pediatrician
	❑ General Check-ups
	❑ Monitor growth and development (Failure to Thrive at Birth)
	❑ Monitor weight
	❑ Feeding evaluation w/Swallow Function Study to assess for Oropharyngeal Dysphagia (referral to Gastroenterologist)
	❑ Growth Hormone Evaluation- Growth Assessment**
	  ❑ Short stature assessment
	  ❑ Bone Age
	  ❑ IGF-1
	  ❑ IGFBP-3
	❑ Evaluate Musculoskeletal (hypermobility, clubbed feet and *pes planus*) Referral to Orthopedic or Pediatric Physical Medicine and Rehabilitation
	❑ Monitor for Scoliosis and Kyphosis – Radiographic survey- (x-ray of spine) if suspected
	❑ Monitor blood work
	  ❑ Glucose (Hypoglycemia concerns)
	  ❑ Vit. D 25 Hydroxy
	  ❑ Calcium Level
	  ❑ B12
	  ❑ RBC (iron and anemia concerns)
	  ❑ Thyroid Function Panel
	  ❑ Parathyroid Level
	❑ Irrigate ears of wax / glue ear (due to rapid buildup of very thick and sticky wax) as needed
	❑ Initiate referrals to sub-specialist
	
	Note: the duplication alters the weight management region of the frontal lobe, causing low birth weight, and slow weight gain is seen until age 5-6, with some remaining skinny into adult years)
		At time of diagnosis and as needed. Assess growth parameters and development at each visit.

Teens and Adults Years Primary Care Physician	❑ General Check-ups ❑ Monitor Weight ❑ Irrigate ears of wax (Glue ear) ❑ Monitor bloodwork (see list above in Pediatrics)	

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	❑ Bone Density- DEXA Scan (after puberty and adults) ❑ NTX Urine Test and CTX Blood Test to evaluate bone calcium absorption and turnover ❑ Monitor for Scoliosis and Kyphosis- Radiographic survey ❑ Evaluate for puberty (referral to Endocrinologist) ❑ Observe for UTI's and Kidney Stones (if suspected) ❑ Initiate referrals to sub-specialists	
Development /Psychiatric clinic	❑ Neurocognitive (developmental) and behavioral evaluation ❑ Clinical screening for developmental delay, atypical behavior, inattention, hyperactivity, anxiety expressing as ADHD, and ASD (Autism Diagnostic Observation Schedule™-ADOS™ to aid in diagnosis of ASD) *ASD = autism spectrum disorder; DD = Developmental Delay*	At time of diagnosis and annually in childhood or as needed
Early childhood intervention	❑ Physical, occupational, and speech therapy evaluations ❑ ABA Therapy Evaluation (if needed)	At time of diagnosis, then as needed.
Psychiatric	❑ Evaluation and management for anxiety disorder ❑ Evaluation and management for depression	Adolescents and Adulthood as needed
Geneticist	❑ Confirmation of diagnosis and periodic follow-up evaluations	At time of diagnosis
Physical Medicine and Rehabilitation (Pediatric and Adult Physiatrist)	❑ Evaluation for motor skills ❑ Evaluation for low muscle tone ❑ Evaluation for hypermobility ❑ Physical Therapy and Occupational Therapy assessment and management (as needed)	As needed
Audiology	❑ Routine audiology evaluation *Note:* they tend to have narrow ear canals, canals that set at an unusual angle, and rapid buildup of earwax	At time of diagnosis, then as needed.
Cardiology	❑ Echocardiogram and ECG/EKG to evaluate for congenital heart defects and aortic root dilatation (enlargement of the aorta). ❑ Electrocardiogram to evaluate for electrocardiographic abnormalities (ie. Prolonged QT) ❑ Evaluate for Dysrhythmias (in teen and adult years)	At time of diagnosis. If normal, repeat echo every 2-3 years*

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Dental	❑ Evaluate for enamel problems, malocclusion, dental crowding, early tooth loss	As needed
Ear, Nose, and Throat (ENT)	❑ Evaluate for palate abnormalities (submucous cleft palate) ❑ Evaluate hearing ❑ Evaluate earwax buildup and removal ❑ Evaluate for swallow dysfunction (Dysphagia, delay in swallow)	As needed
Gastroenterology / Feeding and Nutrition	❑ Feeding assessment for failure to thrive growth retardation ❑ Evaluation for Feeding Therapy (in childhood) ❑ Evaluation for short term feeding tube, if needed for nutrition and growth in children. Referral for Feeding Therapy. ❑ Assess for Oropharyngeal Dysphagia ❑ Evaluate for reflux ❑ Evaluate for Gastroparesis ❑ Evaluate for constipation ❑ Evaluate for low digestive enzymes (if suspected) ❑ Evaluate anxiety level. This has been seen to be a major cause of decrease in desire to eat.	At time of diagnosis, then as regularly as needed.
Neurology	❑ Brain MRI for structural malformations ❑ EEG for seizures ❑ Evaluate for headaches *Note: It has been frequently reported, and observed, that headaches are often been related to conditions related to the syndrome, such as dehydration, poor sleep, anxiety, and overheating. They are often relieved with sleep/nap in the children and adults.* ❑ Referral for sleep study (if sleep apnea is suspected)	At time of diagnosis, then as needed
Ophthalmology	❑ Evaluate for visual acuity deficits and refractive errors ❑ Strabismus ❑ Photophobia ❑ Tracking and convergence ❑ Depth perception dysfunction ❑ Visual 3-D dysfunction ❑ Visual-Cortical Impairment ❑ Referral for vision therapy (if needed)	At time of diagnosis, then annually.

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Speech/language pathology	❑ Swallow function study	At time of diagnosis (if in childhood).
Sleep Study	❑ This depends on age and symptoms. Most children with PTLS do not have frequent awakening, but a fair proportion have obstructive and or central sleep apnea.	At age child will cooperate
Urology	❑ Renal Ultrasound to evaluate for renal abnormalities ^ ❑ UTIs ❑ Kidney Stones (small crystals passed in urine to <5mm have been reported)	At time of diagnosis and as needed
Endocrinology	❑ Growth Hormone Evaluation- Growth Assessment** ❑ Short stature (bone age, IGF-1, IGFBP-3) ❑ Pituitary function ❑ Testosterone, Estrogen, Progesterone (teen and adult years) ❑ Vit. D 25 Hydroxy ❑ Calcium Level ❑ B12 ❑ RBC (iron and anemia concerns) ❑ Platelets (thrombocytes) ❑ Thyroid Function Panel ❑ Parathyroid Level ❑ Phosphates ❑ Digestive Enzymes Amylase and Lipase ❑ Glycemia levels (Hypoglycemia)	Early Childhood and as needed throughout the lifetime
Orthopedic	❑ Scoliosis Evaluation ❑ Kyphosis Evaluation ❑ Bone Density (DEXA Scan) for evaluation of early bone density loss (osteopenia and osteoporosis) ❑ Club foot or foot deformities, *pes planus*	As needed DEXA Scan as early as 16 years, or as recommended by physician

Ref. Original in 2012 written by Dr. L Potocki and Pilar Magoulas as observed in early studies. Update written by Julie Smith-Centeno from updated clinical reports and observations. These are reviewed and recommended by Dr. L. Potocki for PTLS Outreach Foundation, Inc. As new information is learned, this will be updated.
*These recommendations are dependent on the age at the time of the initial evaluation and may be subject to change as we learn more about the cardiac features of individuals with PTLS.
^ Renal ultrasound may not detect reflux in the kidneys; therefore, further evaluation may be warranted.
** See note: Growth Hormone- Evaluate for short stature: bone age, IGF-1, IGFBP-3, referral to endocrinologist.

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