SYNGAP RESEARCH FUND

Collaboration. Transparency. Urgency.

MEDICAL CARE CONSIDERATIONS *for* SYNGAP1 ENCEPHALOPATHY

ICD-10: F78.A1

SEPTEMBER 2021 (v1)

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Patients with SYNGAP1 encephalopathy should be seen for regular wellness checkups and screenings. Inform staff that extra time will be needed for visits to accommodate sensory or motor issues. ***Currently, SYNGAP1 is considered a permanent disability for most patients. There is no specific treatment for SYNGAP1.***

MEDICAL RECORDS

SYNGAP1 families have free access to a Medical Records Solution called Ciitizen via SynGAP Research Fund (SRF), they can sign up at no charge and have all records collected and kept in one place via **Ciitizen.com/SYNGAP1.** Those already enrolled in Ciitizen can share the entire medical record in one step. Alternatively, parents and care-givers should keep a binder of health records. Help them to include: genetic testing results, summaries of all doctor visits (including specialist referrals), summaries of hospital admissions, laboratory studies, EEG, x-ray reports and other imaging results. For questions contact us at info@syngapresearchfund.org.

ICD-10 CODE

Effective October 1, 2021 SYNGAP1 has been assigned the code **F78.A1** in the US. Clinicians should use this code to help improve diagnosis, care, surveillance, research, and access.

MEDICAL DISCLAIMER

All information, content, and material provided in this document is for informational purposes only and is not intended to serve as a substitute for the consultation, diagnosis, and/or medical treatment of a qualified physician or healthcare provider. Always seek the advice of your physician or other qualified health provider with any questions you may have regarding a medical condition.

Areas of Assessment	Assessment Details	Std Age Appropriate Wellness Guidelines	Every 6 Months	Baseline	Every Visit
Genetics/ Testing Results	Counsel family on genetic test results and refer to genetic counselor if appropriate; Family to keep a copy of genetic results.			✓	
General	Update current medications and allergies.				✓
	Weight – Height - Head circumference - Tanner Stage.	✓			
	Laboratory evaluations.	✓			
Neurology	Screen for presence of seizures and spells suspicious for seizures. Record description and frequency of seizures.				✓
EEG	Seizures without abnormal EEG can be present or abnormal EEG without seizures with increased seizure threshold. *Per neurologist or epileptologist.			✓*	✓
Brain MRI	Rule out structural problems (typically normal in SYNGAP1 patients). *Per neurologist or epileptologist.			✓*	✓
Gastro-intestinal	Review: feeding methods, appetite, chewing ability, choking and length of feeding time.				
	Screen for constipation and toilet training.				✓
Urology	Delayed bladder emptying and bladder distension. Toilet training.				✓
Nutrition	Nutrition screening calories, fluids.				✓
Orthopedics/ Rehabilitation	Estimate curvature of spine. Recheck every 6 months if scoliosis is present; Refer to Orthopedics if >20 degrees.	✓	✓		
	Screen for contractures and use or need of devices to prevent them (AFOs and splints.)	✓	✓		
	Screen for needs and use of mobility aids.	✓			

Areas of Assessment	Assessment Details	Std Age Appropriate Wellness Guidelines	Every 6 Months	Baseline	Every Visit
Development	Documentation of baseline gains and losses of milestones. Fine motor: hand use: raking grasp, pincer grasp, rake, holding cup or spoon. Gross motor: sitting, standing, and walking. Language: coo, babble, laugh, words.	✓			
Behavioral	Screen for anxiety and depression such as withdrawal, screaming and irritability. Inquire about sensory processing difficulties. Aggression and OCD.				✓
Sleep	Review sleep initiation, staying asleep, night seizures and frequency of nocturnal interventions by caregivers.				✓
	Review safety of bed and bedroom.	✓			
Screenings	Vision screening that includes acuity, spatial, depth, visual fields and cortical visual impairment. Annual dental health screening.	✓			
Education/ Therapies	Review for presence of current Individualized Educational Plan (IEP). Documentation of therapies (type and frequency.)				✓
Family/Social	Assess for family stress (financial, social, fatigue).				✓
Resources	Review available community, insurance resources (aka DMV permit, respite care, etc.) In adolescent patients review plans for obtaining guardianship. PCP may be required to write Letters of Medical Necessity for equipment and sign school medication forms. *Referral to social work and local government agency for permanent disability application and benefits.			✓*	

GENETICS

For suspicion of SYNGAP1 syndrome or VUS (Variant of Uncertain Significance), referral to a geneticist or genetic counselor is recommended. Contact SRF for guidance at info@syngapresearchfund.org. A VUS may need to have additional testing every few years if there is a high suspicion of SYNGAP1 mutation.

GROWTH, DEVELOPMENT AND NUTRITION

Poor weight gain

Oral aversion is common. Some SYNGAP1 patients have severe problems with nutrition. Consider highly caloric foods (oils, syrups, avocado) and GI and nutrition consults. May need a gastrostomy-tube to maintain growth.

Developmental Milestones

Developmental delay. Therapies to consider are: speech therapy (ST), physical therapy (PT), feeding therapy (FT), occupational therapy (OT), augmentative and alternative communication (AAC), hippotherapy (horse) and swim/pool therapy, music therapy and Applied Behavior Analysis (ABA) therapy.

GASTROENTEROLOGY

Constipation is a very common problem. Laxatives (Miralax, Milk of Magnesia, glycerin suppositories) are often a part of long-term treatment with a goal of one soft BM per day.

UROLOGY

Urine retention: delayed bladder emptying and bladder distension. If present, referral to urology may be needed. Constipation can increase risk of UTIs and time toilet training can be achieved in some cases.

NEUROLOGY

Seizures

Median seizure onset age was 2 years in recent study (Vlaskamp et. al. 2019, Neurology). Refer to Neurology for seizures and spells suspicious for seizures. Neurology follow-up every 6 months if treated with an anticonvulsant. It is difficult to recognize some of the partial seizures in SYNGAP1 and can be EEG negative. EEG at diagnosis is always recommended, long EEGs (days) commonly necessary. Most common seizures: eyelid myoclonia with absences, myoclonic seizures, atypical and typical absences, and atonic seizures. Seizures commonly triggered by eating in 25%.

Abnormal Movements

Hypotonia and/or ataxia of gait abnormalities are common.

ORTHOPEDICS/REHABILITATION

Increased risk of neuromuscular scoliosis and contractures with age. Neurologists or rehabilitation specialists may help to maintain function and prevent contractures.

Equipment and accommodations frequently needed.

GYNECOLOGY

Menarche comes at the usual time. There are no known endocrine abnormalities related to SYNGAP1: Counsel family if seizure frequency corresponds with menstrual cycle. Menses suppression should be considered. The full impact of menses suppression on bone and circulatory health should be discussed and understood; IUD is a consideration (slow-release progestogen, called Levonorgestrel, recommended as preferred method by the American College of Obstetrics and Gynecology (ACOG). Well-woman examinations should include breast exams.

PSYCHOLOGICAL/BEHAVIORAL

Issues with inattention/anxiety. Auditory processing is delayed and may be misinterpreted as disinterest; allow for this delay when assessing non-verbal language by allowing additional time for responses to questions or commands. Behavioral inconsistency is typical and may be affected by physical factors such as sleep or environment. Assess for intolerance of excessive stimuli. Assessment for autism spectrum disorder, autism is common. Psychiatric evaluation later in life may be necessary as there is elevated risk for severe aggression, obsessive compulsive disorder (OCD) and schizophrenia. Early ABA referral even without diagnosis of autism or autism spectrum has been very helpful in many patients.

SLEEP

Disrupted sleep: Circadian rhythm is often disrupted. Patient may be getting out of bed, which could be unsafe if able to wander; consider a tent-style bed or similar engineering controls to keep child in bed and safe (cover by some insurance as Durable Medical Equipment DME). Consider sleep study and otorhinolaryngology (ENT) referral for tonsillectomy evaluation.

PAIN

Pain assessment and sensitivity: Patients may have an atypical pain response, with higher thresholds. Can have serious lesions without significant pain.

SCREENING

Ophthalmology

Difficult vision assessment. Strabismus and cortical visual impairment can be present.

Auditory

Atypical auditory processing can be present. Noise-reduction headphones can be helpful.

Dental

Routine cleanings needed and may require anesthesia.

EDUCATION/THERAPIES:

Most SYNGAP1 patients may have moderate to severe intellectual disability. Individualized Educational Plan (IEP) and therapy challenges: Educators may not have experience with SYNGAP1 syndrome. They should focus on communication, mobility, and socialization with attention to apraxia and neurosensory needs.

SOCIAL CONCERNS

Increased family stress: Family may need respite care. Referral to social work and local government agency for permanent disability application and benefits, **currently SYNGAP1 is considered a permanent disability for most patients.** In adolescent patients review plans for obtaining guardianship and Special Needs Trust. SYNGAP1 is now part of the Social Security Compassionate Allowances Program for People with Severe Disabilities, *Syngap.fund/SSA*.

HOSPITALIZATION CONCERNS

Anesthesia sensitivity, impaired proprioception. May need special accommodations. Patients may need lower doses of anesthetics or analgesics.

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