

CHARGE SYNDROME CHECKLIST: HEALTH SUPERVISION ACROSS THE LIFESPAN (From head to toe)

Revised by K. Blake 2024. www.drkimblake.com

		INFANCY (0-2 years)	CHILDHOOD (3-11 years)	ADOLESCENCE (12-17 years)	ADULTHOOD (18+ years)
GENETICS	Clinical diagnosis (Blake et al. or Verloes or Hale et al. criteria)				
	Genetic testing – Genetics consult (CHD7 analysis, array CGH)				
	Genetic counselling; consider Episign test if genetics is negative				
NEUROLOGY	CNS malformations/hypoplasia olfactory bulb/temporal bone (semicircular canal) malformations – requires MRI/CT				
	Seizures – consider EEG; migraine and abdominal migraine				
	Cranial nerve problems – absent sense of smell, facial nerve palsy, sensorineural hearing loss/balance, swallowing problems				
EYES, EARS, NOSE AND THROAT	Coloboma, Ophthalmology and functional vision assessment; risk of retinal detachment				
	Corneal exposure – lubricating eye drops +/- tarsorrhaphy				
	Photophobia – tinted glasses, sunhat				
	Choanal atresia/tracheoesophageal fistula ENT/Plastics consult				
	Cleft lip and palate/plastics, SLP.				
	Audiometry/tympanometry; monitor for frequent ear infections				
	Adaptive services for individuals with deafness/blindness				
	Cochlear implants/bone bridge implant assessment				
	Obstructive sleep apnea – monitor for tonsil/adenoid hypertrophy				
	Excessive secretions +/- Botox. Glycopyrrolate				
	Dental cleaning under anesthetic, orthodontic treatments				
CARDIOLOGY RESPIROLOGY	Cardiac malformations common – major/minor defects, vascular ring/arrhythmias (x-ray, ECG, Echocardiogram)				
	Postural Orthostatic Tachycardia Syndrome (POTS) vagal stimulation				
	Anesthesia risk of post operative airway obstruction/aspiration, preoperative assessment, documentation. Combine surgical procedures to limit anesthesia				
GASTROENTEROLOGY GENITOURINARY	Gastroesophageal reflux – Gastroenterology consult – consider motility agents with proton pump inhibitor				
	Poor suck, chew, swallow; feeding team +/- gastrostomy +/- jejunostomy				
	Aspiration risk, tracheoesophageal fistula – swallowing studies				
	Constipation consider Senna glycoside +/- polyethylene glycol				
	Bowel obstruction volvulus due to scarring and malrotation				
	Renal anomalies and reflux – abdominal u/s, blood pressure				
MSK	Scoliosis/kyphosis. Physio +/- corset				
	Mobility +/- ID cane (affected by balance, hypotonia)				
IMMUNE SYSTEM	Routine immunizations/antibody titers to immunizations.				
	Recurrent infections – Immunology consult, vitamin D and Zinc.				

CHARGE SYNDROME CHECKLIST: HEALTH SUPERVISION ACROSS THE LIFESPAN (From head to toe)

Revised by K. Blake 2024. www.drkimblake.com

		INFANCY (0-2 years)	CHILDHOOD (3-11 years)	ADOLESCENCE (12-17 years)	ADULTHOOD (18+ years)
ENDOCRINOLOGY	Hypogonadotropic hypogonadism – LH, FSH by 3 months				
	Genital hypoplasia (undescended testes, orchidopexy)				
	Puberty delayed/halted – Endocrinology consult common				
	Osteoporosis – DEXA scan Vit D 2-3,000 daily				
	Poor growth – Endocrinology consult – GH stimulation test				
	Obesity - monitor				
	Fertility, contraception, menstruation challenges - discuss				
DEVELOPMENTAL AND PSYCHOLOGY HEALTH	Gross, fine motor, sensory skills – Physio and OT treatment				
	Total communication – SLP, signing, gestures, AAC				
	Deafblind consult; orientation and mobility training				
	Psychoeducational assessment, Individualized Education Plan				
	CHARGE Non verbal pain assessment (NVPA)				
	Sleep disturbances – consider melatonin/magnesium				
	Behavior management – self regulation, impulse control, anxiety, OCD, emotional and executive dysfunctional issues				
	Life skills/adaptive behavior/social skills/social play				
	Mental health and wellbeing.				
	Sexuality, sexual challenges, and assistance				
	Family stress and sibling services support				
	Transition plan for work and future living				
	Medical self-management – medications, visits, explaining CHARGE syndrome to others. Transitioning to adult services. Seeing healthcare provider independently.				

Resources

Website www.drkimblake.com Pamphlet(s) Presentations/research articles.	International CHARGE Syndrome conference, USA.
The CHARGE Syndrome Foundation (http://chargesyndrome.org/about/charge.asp)	German CHARGE Syndrome Conference
Book – CHARGE Syndrome (Genetic syndromes and Communication Disorders series), 2 nd ed. Hartshorne TS, Hefner M, Blake K, 2021	Deafblind International (http://www.deafblindinternational.org/index.htm)
CHARGE Syndrome Australasia eLearning: https://understandingchargesyndrome.org/www.chargesyndrome.org.au	CHARGE Syndrome Facebook Group and Listserv
CNVPA – Non-vocal pain assessment https://www.chargesyndrome.org/wp-content/uploads/2016/03/non-vocal-pain-assessment.pdf	Open hands, open access: Deaf blind intervener learning modules (Home NCDB Moodle)
Perkins School for the Blind eLearning (http://www.perkinselearning.org/videos)	Feeding scale/pain scale
Website (Feeding scale for CHARGE - Dr Kim Blake) Feeding scale for CHARGE syndrome	OMIM Entry #214800 CHARGE Syndrome (http://www.omim.org/entry/214800)

Abbreviations used in checklist

CGH comparative genomic hybridization	LH luteinizing hormone
CNS central nervous system	MRI magnetic resonance imaging
ENT ear, nose, and throat	MSK musculoskeletal
FSH follicle stimulating hormone	U/S ultrasound
GH growth hormone	CUG voiding cystourethrogram
HRT hormone replacement therapy	OT Occupational Therapy
SLP Speech language therapy	AAC Augmentative Alternative Communication

Professionals who contributed to editing the CHARGE syndrome checklist

Name	Affiliation
Claudia Junghans	CHARGE Syndrome e.v. / Germany
David Brown	Freelance Educational Specialist
Kasee Stratton-Gadke	Mississippi institute on disabilities
Madelene Rich	CHARGE Syndrome Australasia
Tim Hartshorne	Central Michigan University