

# CHARGE SYNDROME CHECKLIST: HEALTH SUPERVISION ACROSS THE LIFESPAN (FROM HEAD TO TOE)

*\*Shaded boxes indicate key assessment points*

		INFANCY (0-2 years)	CHILDHOOD (3-11 years)	ADOLESCENCE (12-17 years)	ADULTHOOD (18+ years)
<b>GENETICS</b>	Clinical diagnosis (Blake et al. or Verloes or Hale et al. criteria)				
	Genetic testing – Genetics consult (CHD7 analysis, array CGH)				
	Genetic counselling				
<b>NEUROLOGY</b>	CNS malformations/hypoplasia olfactory bulb/temporal bone (semi-circular canal) malformations – requires MRI/CT				
	Seizures – more common at older ages – consider EEG				
	Cranial nerve problems – monitor for absent sense of smell, facial nerve palsy, sensorineural hearing loss, vertigo, swallowing problems				
<b>EYES, EARS, NOSE AND THROAT</b>	Coloboma, risk of retinal detachment - Ophthalmology consult (dilated eye exam in infancy, vision assessments)				
	Corneal exposure – lubricating eye drops				
	Photophobia – tinted glasses, sunhat				
	Choanal atresia/cleft palate/tracheoesophageal fistula - ENT/Plastics consult				
	Audiometry and tympanometry, monitor for recurrent ear infections				
	Adaptive services for individuals with deafness/blindness				
	Cochlear implant assessment if applicable				
	Obstructive sleep apnea – monitor for tonsil/adenoid hypertrophy				
	Excessive secretions – consider Botox, medication				
	Dental issues – consider cleaning under anaesthetic				
<b>CARDIOLOGY RESPIROLOGY</b>	Cardiac malformations common – major/minor defects, vascular ring or arrhythmias possible (echocardiogram, chest x-ray, ECG) - Cardiology consult				
	Sinusitis, pneumonia, asthma - monitor				
	Anesthesia risk (difficult intubations/post-op airway obstruction/aspiration) – extensive pre-operative assessment, combine surgical procedures				
<b>GASTROENTEROLOGY GENITOURINARY</b>	Gastroesophageal reflux – Gastroenterology consult – consider motility agents with proton pump inhibitor				
	Poor suck/chew/swallow - feeding team assessment/intervention				
	Aspiration risk, tracheoesophageal fistula – swallowing studies				
	May need supplemental feeds – frequently requires gastrostomy tube or gastrojejunostomy tube				
	Constipation – consider Senna glycoside with polyethylene glycol				
Renal anomalies – abdominal u/s +/- VCUG, blood pressure monitoring					
<b>ENDOCRINOLOGY</b>	Hypogonadotropic hypogonadism – LH, FSH by 3 months				
	Genital hypoplasia (if undescended testes - consider orchidopexy)				
	Delayed puberty – Endocrinology consult - gonadotropin levels, HRT				
	Osteoporosis – DEXA scan				
	Poor growth – Endocrinology consult – GH stimulation test, GH therapy				
	Obesity - monitor				
	Fertility and contraception - discuss				
<b>IMMUNE SYSTEM</b>	Note presence of thymus at open heart surgery				
	Routine immunizations/antibody titres to immunizations in adolescence				
	Recurrent infections – Immunology consult				
<b>MSK</b>	Scoliosis/kyphosis- monitor				
	Mobility (affected by ataxia, hypotonia) - evaluate				
<b>PSYCHOLOGY DEVELOPMENTAL</b>	Assess gross and fine motor skills – Occupational Therapy, Physiotherapy				
	Communication, language, writing abilities – Speech Language Therapy				
	Consider deaf-blind consultant				
	Prepare for transitions to school, situations, places, systems				
	Psychoeducational assessment, Individualized Education Plan				
	Sleep disturbances – consider melatonin				
	Behavior management – self-regulation, impulse control, anxiety, obsessions, compulsions, anger				
	Toileting skills - support				
	Life skills/adaptive behaviour/social skills/social play				
	Address sexuality				
	Family stress – offer supports and resources				
Medical self-management – work on managing medications, understanding conditions, seeing healthcare provider independently					

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## Abbreviations Used in Checklist

CGH - comparative genomic hybridization

CNS - central nervous system

CT - computed tomography

DEXA scan - dual energy X-ray absorptiometry

EEG - electroencephalogram

ENT - ear, nose and throat

FSH - follicle stimulating hormone

GH - growth hormone

HRT - hormone replacement therapy

LH - luteinizing hormone

MRI - magnetic resonance imaging

MSK - musculoskeletal

U/S - ultrasound

VCUG - voiding cystourethrogram

## Resources

- The CHARGE Syndrome Foundation (<http://chargesyndrome.org/about-charge.asp>)
- The CHARGE Informational Pack for Practitioners (SENSE UK) (<https://www.sense.org.uk/content/charge-information-pack-practitioners>)
- Book - CHARGE Syndrome (Genetics and Communication Disorders), 1<sup>st</sup> ed. Hartshorne TS, Hefner M, Davenport S, Thelin J. 2011
- OMIM Entry #214800 CHARGE Syndrome (<http://www.omim.org/entry/214800>)
- CHARGE Syndrome International Conference
- CHARGE Syndrome Listserv
- CHARGE Syndrome Facebook Group
- Perkins School for the Blind e-learning (<http://www.perkinselearning.org/videos>)
- Deafblind International (<http://www.deafblindinternational.org/index.htm>)
- Open hands, open access: deaf-blind intervener learning modules (<http://moodle.nationaldb.org>)

## Key General References

1. Blake K, Prasad C. 2006. CHARGE syndrome. Orphanet J Rare Dis 1: 34
2. Brown D. 2005. CHARGE syndrome “behaviors”: challenges or adaptations? Am J Med Genet Part A 133A: 268-272
3. Hsu P, Ma A, Wilson M, Williams G, Curotta J, Munns CF, Mehr S. 2014. CHARGE syndrome: a review. J Pediatr Child Health 50: 504-511