

# 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 (semicircular 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/postop airway obstruction/aspiration) – extensive preoperative 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 Gastro-jejunostomy 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 deafblind 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 XRay 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/aboutE\\_charge.asp](http://chargesyndrome.org/aboutE_charge.asp))
- The CHARGE Informational Pack for Practitioners (SENSE UK) (<https://www.sense.org.uk/content/chargeEinformationEpackEpractitioners>)
- Book E 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 eLearning

(<http://www.perkinselearning.org/videos>)

- Deafblind International (<http://www.deafblindinternational.org/index.htm>)
- Open hands, open access: deafblind 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: 268E272
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: 504E511