

Appendix 2: Major sentinel congenital anomalies list

System	Congenital Anomaly
Neural	1. Anencephaly
	2. Acrania
	3. Craniorachischisis
	4. Encephalocele
	5. Hydranencephaly
	6. NTD with hydrocephalus
	7. NTD without hydrocephalus
	8. Hydrocephalus
	9. Atresia of foramina of Luschka and Magendie
	10. Dandy-Walker malformation / variant (DWM)
	11. Dandy-walker syndrome
	12. Aqueductal stenosis
	13. Hydrocephalus X-Linked
Cleft Lip / Palate	14. Cleft lip & palate
	15. Cleft lip
Tracheo-Esophageal	16. Esophageal atresia
	17. Tracheoesophageal fistula (TEF)
Small & Large Intestinal	18. Atresia small or large intestine (NOTE: *cannot separate small & large intestinal atresia in BIS).
	19. Duodenal atresia
	20. Abnormal small or large bowel
	21. Double bubble
	22. Bowel obstruction small or large intestine
	23. Stenosis small or large intestine
	24. Imperforate anus

System	Congenital Anomaly
Hypospadias	25. Hypospadias (NOTE: * BIS does not collect epispadias).
Limb Reductions	26. Limb reduction defect(s) (LRD) - lower limb
	27. Limb reduction defect(s) (LRD) - upper limb
	28. Radial ray anomaly (absent thumb)
	29. Adactyly (absent fingers/ toes)
	30. Ectrodactyly (lobster-claw / cleft hand)
Gastroschisis	31. Gastroschisis
Omphalocele	32. Omphalocele
Renal Agenesis	33. Renal agenesis
Heart	34. Hypoplastic left heart syndrome (HLHS)
	35. Tetralogy of Fallot (TOF)
	36. Transposition of great vessels (TGA)
	37. Transposition of great arteries - congenitally corrected (CCTGA)
Trisomies	38. Trisomy 21 (Down syndrome)
	39. Trisomy 21 (Down syndrome) - translocation
	40. Trisomy 21 (Down syndrome) - mosaic
	41. Trisomy 18
	42. Trisomy 13

Source: adapted from list jointly created by BORN Ontario and the OCAC (Ontario Congenital Anomalies Committee) in 2017.