Table 1: Multisystem features of 22q11.2 deletion syndrome (used with permission, Basset et al 2011)

Common features[*]	Relevant age groups			Selected rarer features[†]	Management		Specialties commonly involved (in addition to family medicine, paediatrics, general internal medicine, radiology)
	Prenatal	Infant to child	Teen to adult		Standard[‡]	Special considerations or attention	
General genetics ▪ Dysmorphic features (>90% of cases)[§] ▪ Multiple congenital anomalies ▪ Learning disability/mental retardation/developmental delay (90%) ▪ Poly-hydramnios (16%)	✓	✓	✓	▪ Fetal loss or infant death	✓	▪ Genetic counselling ▪ Medical management ▪ Gynaecological and contraceptive services	▪ Medical genetics ▪ Obstetrics and gynaecology
Cardiovascular (conotruncal/other) ▪ Any congenital defect (including minor) (50%-75%) ▪ Requiring surgery (30%-40%)	✓	✓	✓	▪ Vascular ring ▪ Dilated aortic root ▪ Arrhythmias	✓	▪ Echocardiogram ▪ Irradiated blood products for infant surgeries ▪ Calcium level	▪ Cardiovascular surgery ▪ Cardiology
Palatal and related (75%) ▪ Hypernasal speech (crying) and/or nasal regurgitation (>90%) ▪ Velopharyngeal insufficiency ± submucous cleft palate (overt cleft palate/cleft lip is less common) ▪ Chronic and/or secretory otitis media ▪ Sensorineural and/or conductive hearing loss (30%–50%)	✓	✓	✓	▪ Laryngeal web ▪ Tracheo-esophageal fistula ▪ Esophageal atresia ▪ Preauricular tags/pits[**] ▪ Microtia/anotia[**]	✓	▪ Speech therapy ▪ Palatal surgery	▪ Speech pathology ▪ Plastic surgery/Cleft palate team ▪ Otorhinolaryngology ▪ Audiology
Immune-related[¶] ▪ Recurrent infections (35%–40%) ▪ T-cells low and/or impaired function ▪ Autoimmune diseases		✓	✓	▪ Immunoglobulin A deficiency ▪ Severe immunodeficiency (0.5%–1%)	✓	▪ Influenza vaccinations ▪ Special protocol[¶] for infants	▪ Immunology ▪ Rheumatology ▪ Otolaryngology ▪ Allergy ▪ Respirology
Endocrine ▪ Hypocalcemia and/or hypoparathyroidism (>60%) ▪ Hypothyroidism (20%), hyperthyroidism (5%) ▪ Obesity (35%, adults)		✓	✓	▪ Growth hormone deficiency ▪ Type 2 diabetes	✓	▪ Vitamin D and calcium supplementation[‖] ▪ Growth hormone ▪ Dietary/exercise counselling	▪ Endocrinology ▪ Dietician

Common features*	Relevant age groups			Selected rarer features†	Management		Specialties commonly involved (in addition to family medicine, paediatrics, general internal medicine, radiology)
	Prenatal	Infant to child	Teen to adult		Standard‡	Special considerations or attention	
Gastroenterological ▪ Gastro-esophageal reflux ▪ Dysmotility/dysphagia (35%) ▪ Constipation ▪ Cholelithiasis (20%) ▪ Umbilical/inguinal hernia	✓	✓	✓	▪ Aspiration ▪ Imperforate anus ▪ Intestinal malrotation ▪ Hirschsprung's ▪ Diaphragmatic hernia	✓	▪ Tube feeding ▪ (Gastrostomy Nissen)	▪ Gastroenterology ▪ General surgery ▪ Feeding team ▪ Respirology
Genitourinary ▪ Structural urinary tract anomaly (31%) ▪ Dysfunctional voiding (11%) ▪ Unilateral renal agenesis (10%) ▪ Multi-cystic dysplastic kidneys (10%)	✓	✓	✓	▪ Echogenic/ hypoplastic kidneys ▪ Duplex kidney ▪ Hydronephrosis ▪ Hypospadias ▪ Cryptorchidism ▪ Absent uterus ▪ Nephrocalcinosis	✓	▪ Ultrasound ▪ Transplant	▪ Urology ▪ Nephrology ▪ Gynaecology ▪ Radiology
Ophthalmology ▪ Strabismus (15%) ▪ Refractory errors ▪ Posterior embyrotoxon, tortuous retinal vessels**		✓		▪ Sclerocornea ▪ Coloboma ▪ Ptosis	✓	▪ Eye exam	▪ Ophthalmology
Skeletal ▪ Scoliosis (45%; 6% requiring surgery) ▪ Cervical spine anomalies/thoracic butterfly vertebrae ▪ Idiopathic leg pains in childhood ▪ Sacral sinus	✓	✓	✓	▪ Cervical cord compression ▪ Craniosynostosis ▪ Upper/lower extremity pre and post axial polydactyly	✓	▪ Radiographs ▪ Orthotics	▪ Orthopaedics ▪ Neurosurgery ▪ Radiology ▪ General surgery ▪ Hand surgery ▪ Physiotherapy
Hematology/Oncology ▪ Thrombocytopenia (30%) ▪ Splenomegaly (10%)		✓	✓	▪ Idiopathic thrombocytopenia ▪ Bernard-Soulier ▪ Autoimmune neutropenia ▪ Leukemia, lymphoma, hepatoblastoma	✓	▪ Surveillance	

Common features*	Relevant age groups			Selected rarer features†	Management		Specialties commonly involved (in addition to family medicine, paediatrics, general internal medicine, radiology)
	Prenatal	Infant to child	Teen to adult		Standard‡	Special considerations or attention	
Neurologic ▪ Recurrent (often hypocalcemic) seizures (40%, adults) ▪ Unprovoked epilepsy (5%)		✓	✓	▪ Polymicrogyria ▪ Cerebellar abnormalities ▪ Neural tube defects ▪ Abdominal migraines	✓	▪ Calcium, magnesium levels ▪ Electroencephalogram ▪ Magnetic resonance imaging	▪ Neurology
Growth and development ▪ Failure to thrive ▪ Motor and/or speech delays (>90%) ▪ Learning disabilities (>90%); mental retardation (~35%) ▪ Short stature (20%)	✓	✓	✓		✓	▪ Early intervention ▪ Sign language ▪ Educational supports ▪ Vocational counselling	▪ Developmental paediatrics ▪ Speech language pathology ▪ Occupational/physical therapy ▪ Neuropsychology ▪ Educational psychology
Neuropsychiatric disorders ▪ Psychiatric disorders (60%, adults) ▪ Childhood disorders (eg, attention-deficit, autism spectrum disorders) ▪ Anxiety and depressive disorders ▪ Schizophrenia and other psychotic disorders (>20%)		✓	✓		✓	▪ Surveillance ▪ Standard treatments	▪ Psychiatry ▪ Developmental pediatrics
Other ▪ Non-infectious respiratory disease (10–20%) ▪ Seborrhea or dermatitis (35%); severe acne (25%) ▪ Patellar dislocation (10%) ▪ Dental problems—enamel hypoplasia/chronic caries ▪ Varicose veins (10%		✓	✓		✓		▪ Respirology/Pulmonary/ Anaesthesia ▪ Dermatology ▪ Rheumatology ▪ Orthopaedics ▪ Dentistry ▪ Vascular surgery

*Rates are estimates only of lifetime prevalence of features for 22q11DS and will vary depending on how cases are ascertained and age of the patient. Features included have prevalence >1% in 22q11DS and significantly higher than general population estimates.
†A selected (and to some extent arbitrary) set of rarer features of note in 22q11DS, emphasizing patients needing active treatment.
‡Standard surveillance, investigations, and management according to involved condition(s).
§Characteristic facial features include long narrow face, malar flatness, hooded eyelids, tubular nose with bulbous tip, hypoplastic alae nasae, nasal dimple or crease, small mouth, small protuberant ears with thick overfolded/crumpled helices, and asymmetric crying facies.
¶Infants only: minimize infectious exposures; initially withhold live vaccines; cytomegalovirus-negative irradiated blood products; influenza vaccinations; respiratory syncytial virus prophylaxis.
‖All patients should have vitamin D supplementation; patients with documented hypocalcemia, relative or absolute hypoparathyroidism, or both may have to have prescribed hormonal forms (eg, calcitriol) supervised by endocrinologist.
**May be important for diagnostic purposes.

Citation: Bassett, A. S., McDonald-McGinn, D. M., Devriendt, K., Digilio, M. C., Goldenberg, P., Habel, A., … The International 22q11.2 Deletion Syndrome Consortium. (2011). Practical Guidelines for Managing Patients with 22q11.2 Deletion Syndrome. *The Journal of Pediatrics*, *159*(2), 332–9.e1. doi:10.1016/j.jpeds.2011.02.039