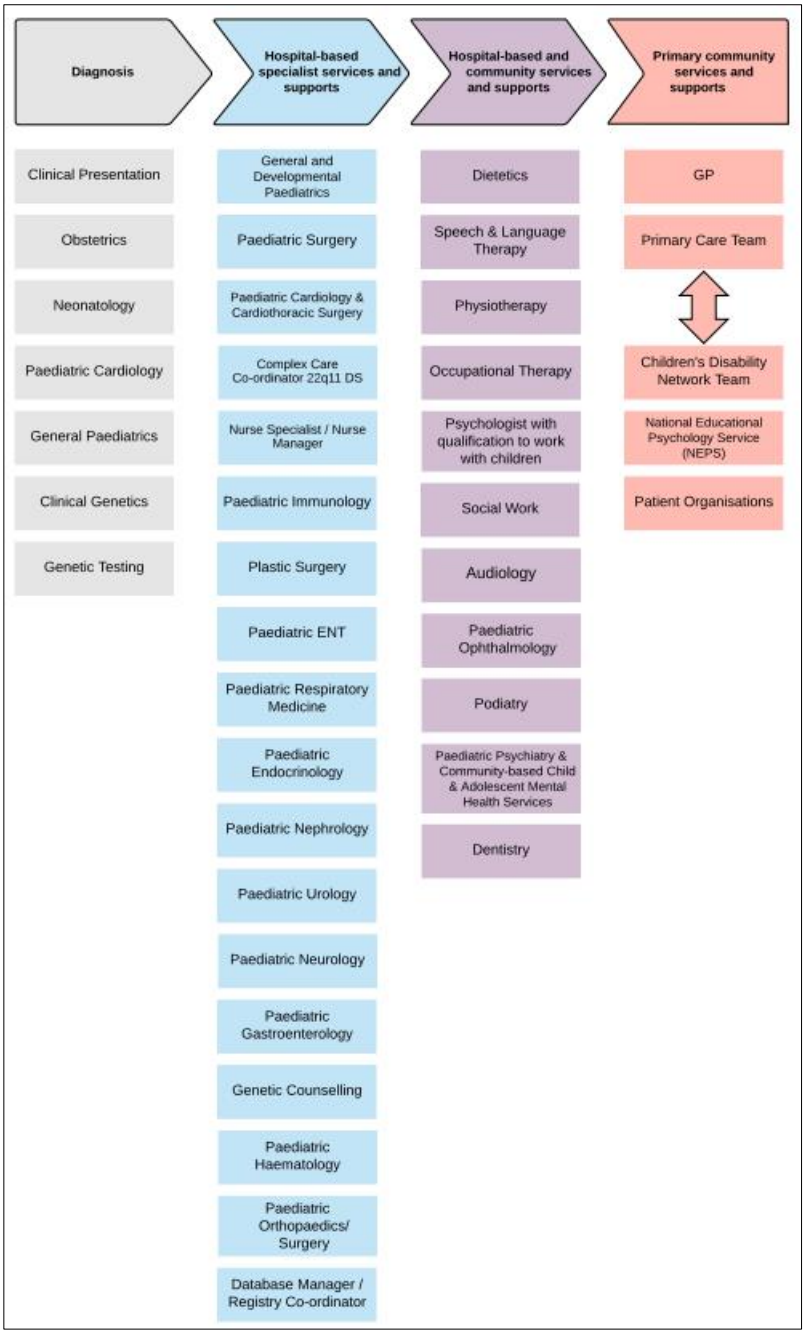
# 22q11 Deletion Syndrome

# Paediatric Care Pathway

HSE National Rare Diseases Office

# 22q11 Deletion Syndrome (DS) Paediatric Care Pathway




## Clinical Characteristics:

**Orphacode: 567**

**Disease Definition:**
22q11.2 deletion syndrome (DS) is a chromosomal anomaly which causes a congenital malformation disorder whose common features include cardiac defects, palatal anomalies, facial dysmorphism, developmental delay and immune deficiency

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| Diagnosis | Clinical Presentation | Family History of 22q11 Deletion Syndrome<br>Congenital heart defects– conotruncal malformations such as truncus arteriosus, tetralogy of Fallot and ventricular septal defect<br>Palatal anomalies - overt cleft palate, cleft lip and palate, velopharyngeal incompetence may lead to hyper-nasal speech, feeding and swallowing difficulties<br>Developmental delay (frequent)<br>Mild facial dysmorphism (frequent) - malar flatness, ptosis, hypertelorism, epicanthal folds, prominent nasal root and vertebral anomalies (butterfly vertebrae, hemivertebrae)<br>Increased frequency of viral infections<br>Immune deficiency - thymic aplasia/ hypoplasia<br>Increased risk of developing an autoimmune condition such as idiopathic thrombocytopenic purpura and juvenile idiopathic arthritis<br>Neonatal hypocalcaemia - usually resolves but can reappear at any age or after an infection, surgery or pregnancy<br>Gastrointestinal anomalies - intestinal malrotation, imperforate anus<br>Hearing loss - increases with age, or can be congenital due to ear anomalies<br>Renal anomalies e.g, renal agenesis, single kidney, duplication of kidneys<br>Dental anomalies - enamel hypoplasia, caries<br>Learning problems and/or psychiatric conditions - attention deficit hyperactivity disorder, schizophrenia |
| Diagnosis | Obstetrics | Abnormal antenatal scan-detection of congenital heart disease, polyhydramnios, single kidney |
| Diagnosis | Neonatology | Diagnosis suspected on clinical examination and detection of anomalies e.g. cardiac defects by ECG or echocardiography, vertebral anomalies by cervical spine X-rays, hypocalcaemia, etc. |
| Diagnosis | Paediatric Cardiology | Cardiac anomalies e.g. truncus arteriosus, tetralogy of Fallot, ventricular septal defect |
| Diagnosis | General Paediatrics | Developmental delay - may present via Public Health Nurse, GP, primary care, educational setting, primary care or occasionally Children's Disability Network Team (CDNT) |

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| | | Referral for diagnostic confirmation may come via Health and Social care professional, Plastic Surgery, ENT, Dental, Endocrinology, Psychiatry, Immunology |
| Diagnosis | Clinical Genetics | Congenital anomalies, developmental delay, dysmorphism |
| Diagnosis | Genetic Testing | Diagnosis confirmed by detection of 22q11.2 deletion - most commonly using microarray CGH |
| Hospital-based specialist services and supports | General and Developmental Paediatrics | Developmental assessment children < 5years<br>Growth monitoring<br>Advice re: sleeping issues<br>Advice re: feeding difficulties<br>Advice re: continence issues<br>Screen for scoliosis (45%) at diagnosis, pre-school and adolescence<br>Renal ultrasound for urinary tract anomalies<br>Investigation for hypocalcaemia and/or hypoparathyroidism, Ionised calcium, TSH, PTH yearly<br>Vitamin D and calcium supplementation<br>Screen for auto-immune endocrine disease<br>Screen for psychiatric condition and refer if indicated<br>EEG (if indicated never as a screen) – epilepsy (5%)<br>Arrange referrals to specialities, and Health and Social Care Professionals as indicated |
| Hospital-based specialist services and supports | Paediatric Surgery | Repair of congenital anomalies, hernia, insertion of percutaneous endoscopic gastrostomy (PEG) |
| Hospital-based specialist services and supports | Paediatric Cardiology & Cardiothoracic Surgery | Manage and treat congenital heart disease -<br>ECG - arrhythmia<br>ECHO for congenital anomaly e.g. Tetralogy of Fallot/ vascular ring, dilated aortic root<br>CT angiogram pre-cardiac surgery |
| Hospital-based specialist services and supports | Complex care co-ordinator 22q11 DS | Support in navigation of health & social care system, patient & family education, transition support, central point of contact, outpatient clinic co-ordination and management, Liaison with stakeholder agencies, advocacy, research, development of Model of Care, outcome measures, advice re global developmental issues |

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| Hospital-based specialist services and supports | Nurse Specialist / Nurse Manager | Expert advice, support and education for patients and carers<br>Point of contact to co-ordinate care and services between hospital and community<br>Co-ordination of care within the hospital<br>Support for child and family especially during in-patient admissions<br>Support and guidance in conjunction with social workers re: P-CAT (Paediatric Community Assessment Tool) respite options and home care supports |
| Hospital-based specialist services and supports | Paediatric Immunology | Immunodeficiency screening<br>Advice re: vaccinations and prophylactic antibiotics |
| Hospital-based specialist services and supports | Plastic Surgery | Assessment and reconstruction e.g. cleft lip and palate, submucosal cleft palate, velopharyngeal abnormalities, craniofacial abnormalities, skin tags |
| Hospital-based specialist services and supports | Paediatric ENT | Investigation for tracheosophageal (TE) fistula, oesophageal atresia, laryngeal web, supraglottic stenosis, microtia/ anotia, recurrent otitis media |
| Hospital-based specialist services and supports | Paediatric Respiratory Medicine | Tracheostomy/ ventilation, obstructive sleep apnoea - CPAP/ sleep studies, recurrent respiratory infections |
| Hospital-based specialist services and supports | Paediatric Endocrinology | Manage faltering growth - growth hormone treatment<br>Manage hypocalcaemia and/or hypoparathyroidism<br>Manage auto-immune endocrine disease |
| Hospital-based specialist services and supports | Paediatric Nephrology | Manage renal issues e.g. renal agenesis, single kidney, multiple cysts |
| Hospital-based specialist services and supports | Paediatric Urology | Renal surgery, if indicated |
| Hospital-based specialist services and supports | Paediatric Neurology | Manage epilepsy |
| Hospital-based specialist services and supports | Paediatric Gastroenterology | Gut failure management |

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| Hospital-based specialist services and supports | Genetic Counselling | All families require referral for genetic counselling at diagnosis<br>Genetic Counselling for young adults with 22q11 at transition<br>No inpatient service, service is out-patient by referral only following genetic diagnosis<br>Urgent referral considered if antenatal diagnosis<br>Review genetic test results<br>Parental microarray follow-up, as appropriate<br>Identify at-risk relatives<br>Cascade testing, as appropriate<br>Autosomal dominant inheritance<br>90% cases are *de novo* with low recurrence risk due to germline mosaicism<br>50% risk for offspring of affected individuals<br>Discuss all available reproductive options |
| Hospital-based specialist services and supports | Paediatric Haematology | Pancytopenia, thrombocytopenia, leukaemia |
| Hospital-based specialist services and supports | Paediatric Orthopaedics/ Surgery | Scoliosis management - bracing, surgery<br>Orthopaedic management of patellar dislocation, foot issues |
| Hospital-based services and supports | Database Manager/Registry Co-ordinator | Create and maintain database of patients attending service<br>Record patient biographical, clinical and research data<br>Audit and quality improvement<br>Ensure minimum data set standards for ERN registry |
| Hospital-based and community services and supports | Dietetics | Nutritional advice in new-born period for congenital anomalies<br><br>Nutrition assessment including monitoring of growth to ensure nutritional adequacy<br><br>Supporting breast milk feeding and fortification of expressed breast milk<br><br>Provision of nutritional support including enteral tube feeding and parenteral nutrition as indicated<br><br>Aid in management of gut failure and manage other co-morbidities with specialist teams outlined below<br><br>Provide nutritional advice to support management of faltering growth, anaemia and hypocalcaemia |

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| | | Assist in the management of feeding and food related difficulties including texture dietary advice if swallowing impairment. Multidisciplinary team working to promote oral development skills and improve dietary intake<br><br>Prevent under-nutrition /aid in management of overweight and obesity |
| Hospital-based and community services and supports | Speech & Language Therapy | Assess and support speech, language and communication needs (SLCN) and / or feeding, eating, drinking & swallowing (FEDS) skills based on individual needs / priorities<br>Initial assessment for feeding difficulties usually hospital inpatient, investigation needs to be linked with hospital based SLT, may need video-fluoroscopy and velopharyngeal insufficiency (palatal assessment FEDS Specialised Services out-patient clinic)<br>Speech delay and rehabilitation<br>Augmentative communication<br>Behavioural support<br>Patient advocacy, patient / staff education, training and awareness<br>Specialist SLT as part of cleft team (if required)<br>Specialist SLT care for FEDS and SLCN in case of tracheostomy |
| Hospital-based and community services and supports | Physiotherapy | Respiratory - respiratory disease (Granulomatous lymphocytic interstitial lung disease – GLILD)<br><br>Pulmonary Rehab<br>Laryngeal and/or tracheal disorders, Aspiration pneumonia (secondary to spinal deformity):<br><br>Chest physiotherapy to address respiratory muscle weakness, dysfunctional breathing and retained secretions, Management of (exertional) dyspnoea<br><br>Obstructive sleep apnoea (post pharyngoplasty): Non-invasive Ventilation<br><br>Neurology - Motor developmental delay, non-progressive dyspraxia:<br><br>Balance and gait training, strength training, promote normal patterns of movement, fine motor control, |

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| | | postural correction, supportive devices, adaptive equipment<br><br>Skeletal - Vertebral malformations/Scoliosis, Talipes Equinovarus, Sprengel's shoulder, Patellar dislocation, Ligamentous laxity (Pes Planus), Equinus, Juvenille Idiopathic Arthritis/Rheumatoid Arthritis, Idiopathic leg pains:<br><br>Rehabilitation post-surgical correction, Provision of brace/orthotics, Gait training – proprioception training, Provision of mobility aids, Stretching and strength training<br><br>Promotion of exercise therapy / exercise counselling for weight management |
| Hospital-based and community services and supports | Occupational Therapy | Fine motor skills and self-management skills for activities of daily living<br>Gross and fine motor support, sensory integration, school liaison, specialised equipment seating/mobility aids/feeding/safety equipment<br>Sleep, behaviour, parenting support, alleviate anxiety |
| Hospital-based and community services and supports | Psychologist with qualification to work with Children | Cognitive/ IQ Assessment - educational assessments pre-school, mid-school, transition to secondary, pre leaving school-vocational planning)<br>Educational needs/ learning supports<br>School placements<br>Autism Spectrum Disorders (ASD) Assessment<br>Support for chronic condition and anxiety management<br>Behavioural support<br>Developmental delay assessment<br>Ideally hospital-based liaison service |
| Hospital-based and community services and supports | Social Work | Psychosocial support: Assess social and family supports, safeguarding<br>Link with community supports as required - GP, Public Health Nurse, Primary Care SW, Local authority SW, Mental Health SW, Disability SW, TUSLA<br><br>Offer 1-1 counselling or GP referral to Counselling in primary care (CIPC)<br>www.hse.ie/eng/services/list/4/mental-health-services/counsellingpc/ |

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| | | Financial support (as required): Patient advocacy, support applications for Medical card, Disability allowance, Supplementary Welfare allowance, Exceptional Needs payment, Long-term illness card, direct to Citizens' information www.citizensinformation.ie/<br><br>Housing and/or mobility issues: Advocacy and support<br><br>Home Care Packages: if issues with activities of daily living - arrange application for inpatients, liaise with Public Health Nurse to arrange for out-patients<br><br>Respite Care: liaise with public health nurse or community disability services to arrange<br><br>Employment issues: Link to Intreo public employment and EmployAbility services www.gov.ie/en/campaigns/fb84c0-intreo/ www.gov.ie/en/service/8578c4-access-the-employability-service/ |
| Hospital-based and community services and supports | Audiology | New-born hearing screen - all children<br>Hearing assessment at diagnosis and then 4-5 yearly<br>Hearing Aids – community-based service |
| Hospital-based and community services and supports | Paediatric Ophthalmology | Hospital-based eye exam following diagnosis<br>Community screen for strabismus, refractory errors (2 yearly in childhood) |
| Hospital-based and community services and supports | Podiatry | Orthotics |
| Hospital-based and community services and supports | Paediatric Psychiatry & Community-based Child & Adolescent Mental Health Services (CAMHS) | Inpatient assessment of acute psychosis, inpatient liaison child psychiatry service – link to local CAMHS, as necessary<br><br>Hospital-based liaison psychiatry service for out-patients<br><br>Baseline psychiatric assessment at diagnosis and as needed with periodic screening throughout childhood and adolescence |

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| | | Community-based screening, assessment and treatment for Schizophrenia, Attention Deficit Hyperactivity Disorder (ADHD), Depression, Anxiety |
| Hospital-based and community services and supports | Dentistry | Initial hospital-based assessment<br>Need for treatment under General Anaesthetic in Hospital/ Community<br>Dental assessment/ Oral Hygienist: Baseline and twice yearly as adult<br>3 monthly oral hygiene visits in childhood |
| Primary and community services and supports | GP | Management of inter-current conditions and acute care<br>Access to local primary care team<br>Co-ordination of local services and supports<br>Refer/communication across services |
| Primary and community services and supports | Primary care team | Children with 22q11 deletion syndrome may have difficulties or delays in development. Some can have their needs met by their local Primary Care services |
| Primary and community services and supports | Children's Disability Network team | Children with 22q11 deletion syndrome who have more complex needs should be referred to their local Children's Disability Network Team (Find a children's disability network team - HSE.ie ) in accordance with the National Access Policy (Introduction (hse.ie) ) using the attached Referral forms (Get a referral for a children's disability network team - HSE.ie) |
| Primary and community services and supports | National Educational Psychology service (NEPS) | School referral to National Educational Psychological Service (NEPS)<br>Developmental Delay Assessment<br>Cognitive / IQ assessment<br>Educational Needs / Learning Support<br>School Placements<br>Link with Children's Disability Network Team (CDNT) psychology |
| Primary and community services and supports | Patient organisations | Advocacy, support and information:<br>22q11 Ireland (www.22q11ireland.org)<br>The International 22q11.2 Foundation (www.22q.org)<br>22q11.2 Society (www.22qsociety.org)<br>Rare Ireland Family Support Network (www.rareireland.ie )<br>Rare Diseases Ireland (www.rdi.ie) |

**Clinical Lead(s):**

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