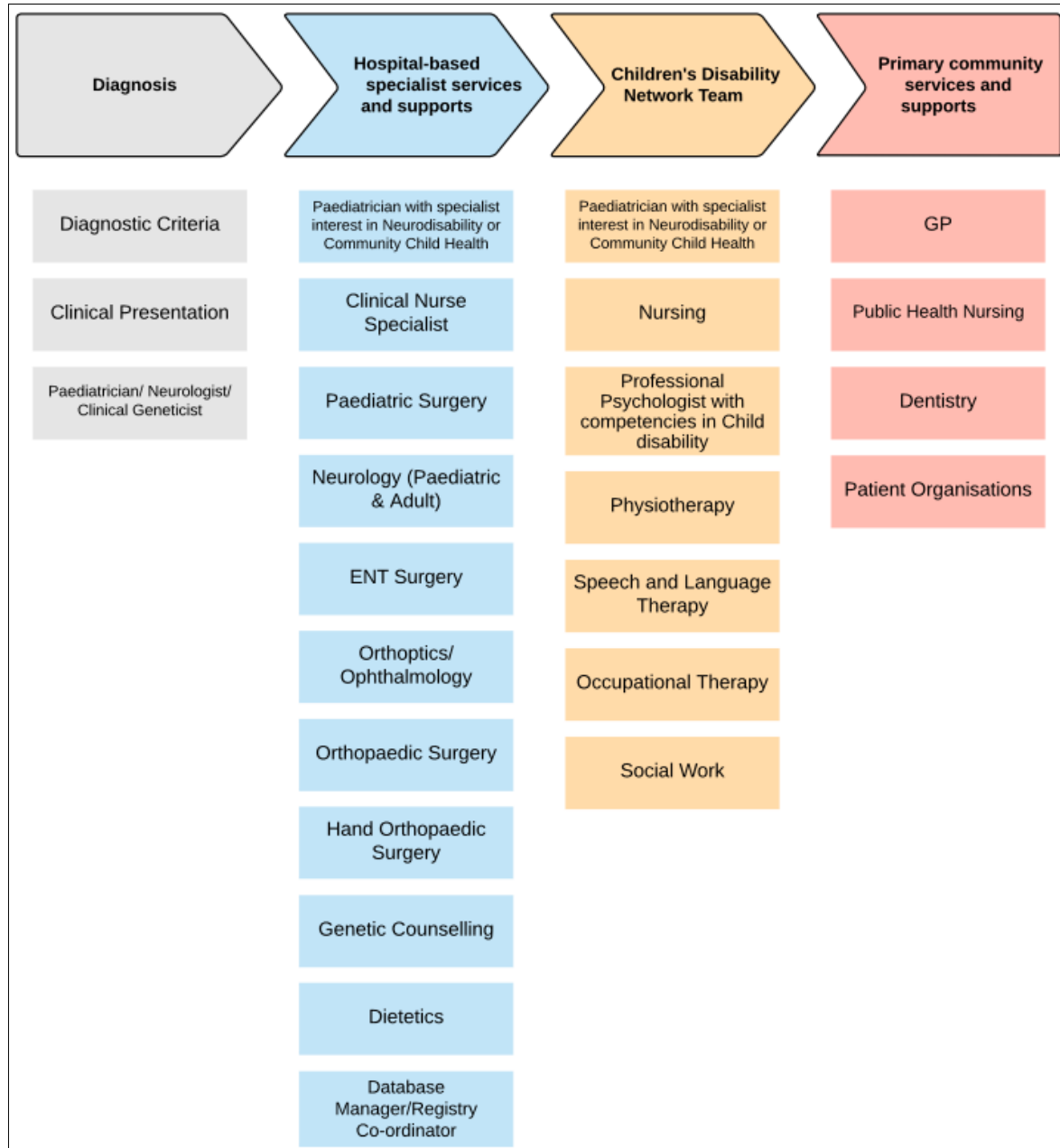




Angelman Syndrome

Paediatric Care Pathway

Angelman Syndrome Paediatric Care Pathway



Clinical Characteristics:

Orphacode: 72

Disease Definition: A neurogenic disorder characterized by severe intellectual disability and distinct facial dysmorphic features

Diagnosis	Diagnostic Criteria	Updated Consensus Diagnostic Criteria (Williams 2005)
Diagnosis	Clinical Presentation	<p>Neonatal to 6 months - poor suck and swallow, feeding difficulties and hypotonia</p> <p>6 months - 2 years developmental delay</p> <p>From 1 year typical features develop - severe developmental delay, absent speech, outbursts of laughter with hand flapping, microcephaly, macrostomia, maxillary hypoplasia, prognathia, neurological problems, gait with uplifted arms, ataxia/ tremor and seizures</p> <p>Other signs include a happy demeanour, hyperactivity, short attention span, excitability and sleeping problems, increased sensitivity to heat, thoracic scoliosis</p>
Diagnosis	Paediatrician/ Neurologist/ Clinical Geneticist	<p>The diagnosis is based on clinical criteria and molecular genetic testing showing deficient expression/ function of the maternally inherited <i>UBE3A</i> allele by:</p> <p>Deletion of the maternal 15q11.2-q13 critical region (60-75%)</p> <p>Paternal uniparental disomy (2-5%)</p> <p>Imprinting defect (2-5%)</p> <p>Pathogenic variant in the <i>UBE3A</i> gene (10%)</p> <p>No genetic defect identified (10%)</p> <p>Characteristic EEG (not associated with seizures), MRI Brain (Normal)</p>
Hospital-based specialist services and supports	Paediatrician with specialist interest in Neurodisability or Community Child Health	<p>Developmental assessment</p> <p>Feeding problems</p> <p>Growth monitoring</p> <p>Scoliosis - annual clinical examination</p> <p>Management of coexisting conditions such as sleep disorder</p> <p>Management of spasticity</p> <p>Advice for behavioural issues – may require multidisciplinary team (MDT) support with speech and language, occupational therapy</p> <p>Hypopigmentation - advise sun block therapy sun protection factor (SPF) > 30</p>
Hospital-based specialist services and supports	Clinical Nurse Specialist	<p>Co-ordination of care from admission to discharge</p> <p>Discharge planning</p> <p>Liaison with Public Health Nurse, Children's Disability Network Team, Complex Need Coordinator, Schools or Day Service</p> <p>Complete Paediatric Community Assessment Tool (PCAT) for respite options and home care supports</p> <p>Co-ordinate clinics/appointments with other discipline specialists</p> <p>Transition Pathway to Adult services</p> <p>Consider Referral to Jack and Jill Children's Foundation (0-6years)</p>

Hospital-based specialist care services and supports	Paediatric Surgery	Percutaneous endoscopic gastrostomy (PEG) tubes and Gastrostomy placement
Hospital-based specialist care services and supports	Neurology (Paediatric & Adult)	Seizure management: Generalised epilepsy - Valproate and Benzodiazepines Cortical myoclonus – Piracetam Atypical seizures - Ethosuximide, Levetiracetam and Benzodiazepines, Refractory epilepsy - steroids, ketogenic diet, vagal nerve stimulation Clinical Trials
Hospital-based specialist services and supports	ENT Surgery	Excessive sialorrhoea (drooling) - botulinum toxins injections, surgery - salivary gland ligation/ re-direction
Hospital-based specialist services and supports	Orthoptics/ Ophthalmology	Strabismus (2 yearly screen) - surgical correction, if required Retinal hypopigmentation/ oculocutaneous albinism – visual evoked potential test (VEPs) Keratoconus - corneal transplant, if severe
Hospital-based specialist services and supports	Orthopaedic Surgery	Thoracic scoliosis (20% childhood, 50% adulthood) - bracing, surgery Consider cardiorespiratory compromise if scoliosis severe Management of spasticity, joint contractures or dislocations
Hospital-based specialist services and supports	Hand Orthopaedic Surgery	Contracture surgery
Hospital-based specialist services and supports	Genetic Counselling	Review genetic testing results – counsel on recurrence risk (from negligible up to 50% depending on underlying mechanism), counsel on reproductive options
Hospital-based specialist services and supports	Dietetics	Nutrition assessment including monitoring of growth to ensure nutritional adequacy (prevent under-nutrition and aid in management of overweight /obesity) Provision of nutrition support including enteral tube feeding Assist in the management of feeding and food related difficulties Provision of texture modified diets if swallowing impairments Education on ketogenic diets and/or low glycaemic diets may be considered in management of seizures
Hospital-based specialist services and supports	Database Manager/Registry Co-ordinator	Create and maintain database of patients attending service Record patient biographical, clinical and research data Audit and quality improvement Ensure minimum data set standards for ERN registry

<p>Children's Disability Network team (CDNT)</p>	<p>Paediatrician with specialist interest in Neurodisability or Community Child Health</p>	<p>Link to multi-disciplinary team assessment Neurodisability CDNT Developmental assessment Feeding problems Growth monitoring Scoliosis - annual clinical examination Management of co-existing conditions such as sleep disorder Management of spasticity Advice for behavioural issues with MDT support from CDNT Hypopigmentation - advise sun block therapy SPF > 30 Safeguarding</p>
<p>Children's Disability Network team (CDNT)</p>	<p>Nursing</p>	<p>Feeding and continence support Education and point of contact for families Liaise with Neurology Nurse Specialist for epilepsy Liaison with patient organisations, MDT team members, hospital professionals, social worker, psychology Transition planning</p>
<p>Children's Disability Network team (CDNT)</p>	<p>Professional Psychologist with competencies in Child Disability</p>	<p>Education and clinical assessment School placement Educational approaches/ intervention Behavioural support - behaviour that challenges, self-injurious behaviour, hyperactivity, impulsivity, aggression Sibling and parent support</p>
<p>Children's Disability Network team (CDNT)</p>	<p>Physiotherapy</p>	<p>Respiratory: Obstructive Sleep Apnoea -Non-invasive Ventilation Neuromusculoskeletal: Movement disorders, Motor developmental delay, Abnormal muscle tone – Spastic diplegia, Ataxic diplegia, Scoliosis, Subluxed or pronated ankles, hip dislocation, Joint contractures and muscle imbalances Promote normal patterns of movement - improve gross motor performance and function Balance, co-ordination and gait & proprioception training. Strength training and stretching. Postural monitoring and management - prevention of contractures and postural correction through bracing, orthotics and taping. Supportive devices and adaptive equipment for non-ambulatory. Rehabilitation post-surgical correction Promotion of ongoing exercise therapy/exercise counselling for weight management (excessive appetite) and to prevent joint contractures and muscle imbalance (through adulthood) Recommendations: Hydrotherapy (love of water), carer-controlled bikes – exercise routine</p>

<p>Children's Disability Network team (CDNT)</p>	<p>Speech and Language Therapy</p>	<p>Support the creation of an 'inclusive communication' environment for all ages, using multi-modal techniques including augmentative and alternative communication aids (AAC) e.g. picture cards or communication boards and signing Continue into adulthood to support communication Assessment and intervention to support Feeding, Eating, Drinking Skills (FEDS) and Speech, Language and Communication Needs (SLCN) Advocacy, education and training for family, staff, educators to support child's participation, activities, learning, independence, and social-emotional well-being across their environments Behavioural support Clinical and / or objective (where appropriate) assessment of swallow safety and efficiency of oral intake in liaison with dietetics, medical and surgical teams</p>
<p>Children's Disability Network team (CDNT)</p>	<p>Occupational Therapy</p>	<p>Assessment and intervention for difficulties participating in activities of daily living; including play, sleep, self-care, fine and gross motor skills Postural management assessment and intervention, including specialist seating and equipment provision Feeding and mealtime support Home and school environmental assessments Assessment for handwriting and assistive technology for school Sensory processing assessment and intervention MDT assessments for developmental assessments</p>
<p>Children's Disability Network team (CDNT)</p>	<p>Social Work</p>	<p>Psychosocial support: Assess social and family supports, safeguarding Link with community supports as required e.g., GP, Public Health Nurse, Primary Care SW, Local authority SW, Mental Health SW, Disability SW, TUSLA Offer 1-1 counselling or GP referral to Counselling in primary care (CIPC) www.hse.ie/eng/services/list/4/mental-health-services/counsellingpc/ 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/ Housing and/or mobility issues: Advocacy and support Home Care Packages: if issues with activities of daily living - arrange application for inpatients, liaise with Public Health Nurse to arrange for out-patients</p>

		<p>Respite Care: liaise with public health nurse or community disability services to arrange</p> <p>Employment issues: Link to Intreo public employment www.gov.ie/en/campaigns/fb84c0-intreo/ Link to EmployAbility services www.gov.ie/en/service/8578c4-access-the-employability-service/</p>
Primary / community services and supports	GP	<p>Management of inter-current conditions and acute care Co-ordinate local services and supports Refer/communicate across services Transition to adult services Primary physician for adults - onward referral to specialist services as necessary</p>
Primary / community services and supports	Public Health Nursing	<p>Community coordinator for complex care Secure equipment Continence support</p>
Primary / community services and supports	Dentistry	<p>Promote good dental hygiene, regular check-ups</p>
Primary / community services and supports	Patient Organisations	<p>Advocacy, support and information: Angelman Syndrome Ireland (www.angelman.ie) Angelman Syndrome Alliance (www.angelmanalliance.org) Rare Ireland Family Support Network (www.rareireland.ie) Rare Diseases Ireland (www.rdi.ie)</p>

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