PAEDIATRIC SERVICE SPECIFICATION

Services for Children and Young People with Down Syndrome

Contents

This service specification has been designed to assist commissioners in the delivery of services for children and young people with Down syndrome.

The specification is not mandatory - it is designed to help implement existing guidelines and should be review and amended to ensure that it meets local needs.

This specification has been developed by the RCPCH in conjunction with the Down Syndrome Association.

Purpose of the Service

Aims of the service

The Down syndrome (DS) service aims to provide optimal child/young person- and family-centred medical care for all children and young people with DS, with the ultimate purpose of improving their physical and mental health, and overall quality of life.

The service also aims to allow children with DS maximise their physical, educational and vocational potential, through improving health outcomes and multi-agency working with educational services.

Key objectives of the service

The objectives of the service are to:

- Improve health outcomes to ensure the full participation of children with DS and their families in their communities

- Provide care that is safe, effective and evidence-based (or based on international consensus on best practice where evidence base is limited)

- Ensure health care services for children with DS are coordinated with the full array of early mental health, educational, social, and other community-based services needed by and provided to children and their families.

- Enhance child development and functional outcomes, and improve family life, through timely screening of complications of DS and the delivery of effective, outcome-based, high-quality medical care.

- Ensure that children with DS receive their care as close to home as possible.

- Ensure effective and seamless transition of care from children's to adult health services where required, and to maximise the young person's understanding of their condition and optimise their autonomy and ability to manage their health care.

- Support parents and families in meeting their responsibilities to nurture and to enhance their children's development.

- Ensure equity of access, quality, consistency, and accountability in the service system by ensuring clear lines of responsibility for the provision of health services to children with DS.

- Promote clinical governance, a quality improvement culture and involvement in research among services for children with DS, in order to assure and continuously improve the quality of care for children with DS.

What is *Down syndrome*?

DS is the commonest chromosomal disorder and the most common identifiable cause of learning difficulty.

DS is associated with complications in multiple organ systems, but the degree of organ involvement varies considerably within the DS population. The commonest of these include:

- Congenital cardiac disease
- Endocrine problems (most commonly hypothyroidism)
- Gastrointestinal problems (including coeliac disease, congenital functional and anatomical bowel obstruction)
- Ophthalmological problems (including refractive error, nystagmus, congenital cataract and glaucoma)
- Otolaryngological problems (including sensorineural hearing impairment, otitis media with effusion, obstructive sleep apnoea)
- Cervical spine disorders
- Haematological malignancy
- Psychological impairment (including psychiatric and behavioural disorders).

As a result of the range of complications with which DS is associated, medical care for children with DS is a multi-disciplinary process led by a paediatrician (who may be a community paediatrician, a paediatrician specialising in neurodisabilities, or a general paediatrician). However, specialist involvement is common due to the frequency of multi-organ complications described above.

Why is managing *Down syndrome* important for improving outcomes?

Life expectancy for children with DS has been steadily rising for the past 5 to 6 decades, and currently stands at around 60 years of age.[1] As a result, early identification and intervention in childhood for medical problems associated with DS will have significant and far-reaching impact upon the overall burden of chronic disease in adults with DS.

[1] Bittles AH, Glasson EJ. Clinical, social, and ethical implications of changing life expectancy in Down syndrome. Developmental Medicine and Child Neurology. 2004;46(4):282-6

National Context

Prevalence

There are approximately 750 babies born with DS every year in the UK, with an incidence of 1:1000 live births. It is estimated that there are currently around 60,000 people with DS in the UK. Screening for DS is part of the national NHS Fetal Anomaly Screening Programme.

Evidence base

There are a number of different models of service provision in current use for children with DS. In some areas there are dedicated specialist DS clinics/services (approximately 15 in the UK, according to a survey undertaken by the Down Syndrome Medical Interest Group UK and Ireland ("DSMIG")), but a similar level of service may also be offered within a more generic child development centre or community paediatric service using local care pathways.

Although the details vary, many of the local care pathways for children with DS are based on national and international standards. Examples include:

- Down Syndrome Medical Interest Group UK and Ireland standards (www.dmsig.org.uk)
- American Academy of Pediatrics Clinical Report: Health Supervision for Children with Down Syndrome (http://pediatrics.aappublications.org/content/early/2011/07/21/peds.2011-1605)
- European Down Syndrome Association Health Care Guidelines for People with Down Syndrome (http://www.edsa.eu/files/essentials/edsa_essentials_2_healthcare.pdf)

Scope

Population covered

Specifically for the purposes of this document, this service is provided for all infants and children (under the age of 19 years) with a clinical diagnosis of DS (regardless of specific karyotype pattern).

Acceptance and exclusion criteria

Acceptance criteria

This service will accept referrals from all providers of health care, but will most commonly accept referrals from neonatal services.

This service will accept referrals up to the child/young person's 19th birthday. Follow up of patients already under the care of the service may occur to a later age, dependent upon local transition arrangements.

Exclusions

This service will not formally accept referrals directly from antenatal screening. Although communication directly from antenatal screening to the service prior to birth is encouraged, referrals in the new-born period will be formally made by neonatal services after birth.

Interdependencies with other services

All services will work directly with, but not limited to, the following professionals to ensure a seamless service:[2]

- Speech and Language Therapy
- Child and Adolescent Mental Health services
- Physiotherapy
- Occupational Therapy
- Medical specialists (as required and depending on local arrangements)
  Including, but not limited to:
  - neonatology
  - paediatric cardiology
  - paediatric ophthalmology
  - paediatric endocrinology
  - paediatric gastroenterology
  - paediatric surgery

[2] Commissioning Safe and Sustainable Specialised Paediatric Services: A Framework of Critical Inter-Dependencies (DH, 2008)

- ENT surgery
- Dermatology
- paediatric orthopaedic/spinal surgery
- paediatric neurology
- paediatric dentistry

The lead paediatrician and local education authority must work in close collaboration to ensure specialist education provision is provided as required for children with DS, including early educational interventions such as portage, and educational psychology.

Service Delivery

Service description/care pathway

The following service standards reflect a *minimum level* of care for all children with DS. Additional services (surveillance, diagnosis or therapy) may be required depending on the needs of individual child with DS.

Core/Essential DS care pathway service standards

1. From diagnosis to transition, all children with Down's syndrome (DS) must be under the care and regular review of a paediatrician with expertise in DS (who may be a community paediatrician, a paediatrician specialising in neurodisabilities, or a general paediatrician).

2. Children with DS are reviewed by the paediatrician regularly in the first year of life. (A frequency of every 3 months is common practice), and subsequently a minimum of once a year.

3. Specialist services to support the lead paediatrician must be commissioned for *all* children with DS, namely:
    - Speech & Language Therapy
    - Paediatric cardiology (echocardiography and clinical review) in the neonatal period
    - Paediatric ophthalmology
    - Paediatric audiology

4. In addition, the following professionals must be commissioned to perform assessment and management of complications of DS, if found necessary after review by the DS specialist:

    - Physiotherapy
    - Medical specialists (as required and depending on local arrangements) including, but not limited to:
        - Paediatric cardiology
        - Paediatric endocrinology
        - Paediatric surgery
        - ENT surgery
        - Dermatology
        - Paediatric gastroenterology
        - Paediatric orthopaedic/spinal surgery
        - Paediatric respiratory specialists / sleep disordered breathing service
        - Paediatric neurology
    - Sexual health service
    - Occupational Therapy
    - Special needs dentistry

- Child and Adolescent Mental Health services

5. The lead paediatrician and local education authority must work in close collaboration to ensure specialist education provision is provided as required for children with DS, including early educational interventions such as portage, and educational psychology.

6. The service must have sufficient clerical and administrative support to facilitate early and sustained communication between clinical specialists, and with children/young people with DS and their families, as well as to capability for audit, governance and service improvement to maximise quality of care.

Additional/Expected standards

i) Neonatal period (or time of diagnosis)

1. Disclosure of clinical suspicion/diagnosis to parents will be made as soon as possible, by a senior clinician (defined as a trained paediatrician, or a senior trainee deemed to be a competent representative of the named paediatrician).

2. Written information regarding Down's syndrome must be made available to parents immediately on disclosure of diagnosis. Examples include:
   - Down Syndrome Medical Interest Group (UK and Ireland Personal Child Health Record ("Red book") insert for children with Down syndrome (http://www.dsmig.org.uk/publications/pchr.html)
   - Early Support: Information for parents: Down syndrome (DfE, 2010) https://www.education.gov.uk/publications/standard/publicationDetail/Page1/ES13
   - Developmental journal for babies and children with Down syndrome (DfE, 2010) https://www.education.gov.uk/publications/standard/publicationdetail/page1/ES49

3. New-born babies with suspected/diagnosed Down's syndrome will have a detailed neonatal examination prior to discharge, with focus on common complications of Down's syndrome, in particular:
   - Cardiac defects
   - Cataracts
   - Bowel atresia/ functional obstruction

4. New-born babies with suspected/diagnosed Down's syndrome must be investigated prior to discharge for common neonatal complications of Down's syndrome, including:
   - Full blood count (to exclude blood disorders related to DS)
   - Electrocardiogram (and echocardiogram if immediately available) (to exclude congenital cardiac anomalies)

- Thyroid function (via routine new-born screening card) (to exclude congenital hypothyroidism)
- Karyotype (to confirm diagnosis)
- Routine neonatal hearing screen (to exclude hearing impairment related to DS)

ii) Specific medical care in childhood

5. Full specifications for surveillance and management of specific medical problems associated with DS are listed in Appendix 1.

iii) Transition

6. The coordinated, multiagency approach outlined above is extended from infancy through to the transition to adult health and social services.

7. Children with DS must have a transition plan in place if appropriate for their needs. As a minimum, formal transition of the care coordinator to the young person's GP must take place. (Care coordination is defined as "a clearly defined function which assures that the objectives and goals agreed with the individual are achieved through the effective delivery of care by the appropriate agency or provider….[and] works best when there is a clearly identified person undertaking this role within a multidisciplinary team." [3])

8. Additionally, it may be necessary to coordinate transition of care to adult learning disability, mental health, or social care services as required.

9. Children who are under regular specialist follow up in a paediatric specialist service must also have a planned transition to an equivalent adult service. Transition must be a planned process, and must follow the Department of Health recommendations.[4]

Applicable national service standards

The service will implement best practice guidelines and quality standards in order to standardise care across local and regional networks. Examples include:

- Down Syndrome Medical Interest Group UK and Ireland (www.dmsig.org.uk)
- American Academy of Pediatrics Clinical Report: Health Supervision for Children with Down Syndrome

[3] Improving care for people with long term health conditions: an "at a glance" guide for healthcare professionals. Information sheet 3: Care coordination. DH, 2011. http://www.dh.gov.uk/prod_consum_dh/groups/dh_digitalassets/documents/digitalasset/dh_124050.pdf

[4] Transition: Getting it Right For Young People (DH, 2006). http://www.dh.gov.uk/en/Publicationsandstatistics/Publications/PublicationsPolicyAndGuidance/DH_4132145

(http://pediatrics.aappublications.org/content/early/2011/07/21/peds.2011-1605)

- European Down Syndrome Association Health Care Guidelines for People with Down Syndrome (http://www.edsa.eu/files/essentials/edsa_essentials_2_healthcare.pdf)

There are also national guidance by medical and health specialists which have particular applicability for children with DS. Examples include:

- Section 1.7: "Management of OME in children with Down's syndrome", NICE clinical guidance 60, *Surgical management of otitis media with effusion*, February 2008 - http://www.nice.org.uk/nicemedia/live/11928/48420/48420.pdf)

Additionally, the following general standards for children's health care are of particular importance for children with DS:

- Children and young people must only receive a service from a provider who takes steps to prevent abuse and does not tolerate any abusive practice should it occur.[5] [6]
- All children and young people who use services must be
    - Fully informed of their care, treatment and support.
    - Able to take part in decision making to the fullest extent that is possible.
    - Asked if they agree for their parents or guardians to be involved in decisions they need to make.[7]

- When children and young people with DS who use paediatric services are moving to access adult services, these must be organised so that all those involved in the care, treatment and support cooperate with the planning and provision to ensure that the services provided continue to be appropriate to the age and needs of the person who uses services.[8]

[5] RCPCH, RCN, RCGP et al (2010) Safeguarding Children and young people Intercollegiate guidance for healthcare staff. http://www.rcn.org.uk/__data/assets/pdf_file/0004/359482/REVISED_Safeguarding_03_12_10.pdf
[6] Care Quality Commission. (2010). *Essential Standards of Quality and Safety*, CQC, London 2010
[7] Ibid.
[8] Transition: Getting it Right For Young People (DH, 2006).

Service Outcomes

- To optimize the quality of life for children and young people with DS through provision of a high quality health care service.
- To ensure the provision of a multi-disciplinary workforce with sufficient capability to identify and manage medical complications in children and young people with DS.
- To ensure children and young people with DS are treated according to evidence-based or best practice guidelines.
- To ensure equitable delivery of care for all children and young people with DS, as close to home as possible
- To ensure coordination of health, education and social care as required for children and young people with DS
- To ensure routine and systematic data collection on performance of each service, and publication of data to allow meaningful comparison among services, ultimately leading to improvement in governance and quality of all local services

Specific indicators for each of the outcomes above will be identified through a consultation period, to include medical experts, families and children and young people with DS and other public representatives. Early suggested indicators include:

- Adherence to national clinical and service guidelines (through audit)
- Workforce data collection
- Surveys of user satisfaction with service, and quality of life, among children and young people with DS and their families
- Indicators of access to services (including time to first consultation, time from referral to specialist assessment)
- Measurement of effectiveness of communication among professionals (within healthcare and with educational/social services)

Appendix 1: Service standards for specific medical problems associated with Down syndrome (DS), from diagnosis to transition:

	First year of life	Early years / pre-school	School Age
Thyroid	All children with DS must undergo the routine newborn blood spot screening test to exclude congenital hypothyroidism.	Thyroid function must be reviewed either: • Annually, on the basis of annual thyroid stimulating hormone blood spot test; or • Biennial serum thyroid function and antibody tests	
Vision	All children with DS must undergo an examination for red reflex to exclude congenital cataract, as part of the routine newborn examination.	By 2 years of age, children with DS must undergo a formal eye and vision test, including squint assessment. All children must also undergo a detailed visual assessment before school age (4 years), to include squint assessment, refraction and acuity.	School aged children with DS must undergo a detailed ophthalmological/optometric assessment a minimum of once every two years.
Hearing	All children with DS must undergo the routine newborn hearing screening test to exclude hearing impairment. Before the child's first birthday, children with DS must undergo a formal audiological review, including hearing assessment & impedance check.	Between one and four years of age, children with DS will undergo an annual audiological review, including hearing assessment & impedance check.	School age children with DS will undergo an audiological review, including hearing assessment & impedance check, a minimum of once every two years.

Breathing	Children with DS must be assessed for symptoms of sleep-related breathing disorder annually until commencing school, with further assessment (including overnight pulse oximetry) arranged where clinically indicated.		School-age children with DS who develop symptoms of sleep-related breathing disorders must be investigated (including overnight pulse oximetry) and managed promptly, including referral to ENT if appropriate.
Heart	By 6 weeks' of age, all children with DS must have a formal cardiological assessment (including echocardiography) to exclude congenital heart disease.	Children with DS must be reviewed annually for signs and symptoms of acquired valvular heart disease, with further assessment (including echocardiography and specialist cardiology referral arranged where clinically indicated.)	
Growth	Children with DS will undergo monitoring of height and weight (plotted on a UK DS-specific growth chart) on an annual basis.		
Haematology	All children with DS will have a blood film assessment in the neonatal period to exclude related blood disorders.		
Gastrointestinal	Assessment (and investigation as required) of common gastrointestinal problems, such as constipation, feeding difficulties and coeliac disease, must take place during each regular medical review.		
Spinal	Assessment (and investigation as required) of developing disorders of the cervical spine must take place during each regular medical review.		