E02/S(HSS)a

NHS STANDARD CONTRACT
FOR BECKWITH-WIEDEMANN SYNDROME WITH MACROGLOSSIA SERVICE (CHILDREN)

PARTICULARS, SCHEDULE 2 – THE SERVICES, A-SERVICE SPECIFICATIONS

Service Specification No.	E02/S(HSS)a
Service	Beckwith-Wiedemann syndrome with macroglossia service (Children)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

1.1 National/local context and evidence base

Beckwith-Wiedemann syndrome (BWS) is a congenital overgrowth syndrome affecting both males and females (Engostrom et al., 1988). Other features of the condition include prenatal and childhood overgrowth (high weight and height), hemi-hypertrophy (asymmetric growth), anterior abdominal wall defects including exomphalos or umbilical hernia, ear lobe creases and posterior ear pits, neonatal hypoglycaemia and predisposition to childhood tumours, particularly Wilms tumour of the kidney (Elliott et al., 1994, Weksberg et al., 2005).

A number of different genetic and epigenetic abnormalities at the imprinted 11p15 growth regulatory region cause BWS and are identifiable in 80% of individuals who fulfil the classical diagnostic criteria (Elliott et al., 1994, Weksberg et al., 2005). The cause in the remainder is not known.

The incidence of BWS is 1 in 13,700 newborns (Engostrom et al., 1988). 50% of this group is estimated to have macroglossia (BWSm) (R. Scott, unpublished data 2010), and therefore there is an incidence of approximately 1 in 28,000 of the target patient group. Based on the number of live births 2009, it is estimated that approximately 22 children with BWSm were born that year.

Children with BWSm are at risk for devastating effects on feeding, drooling, speech, and dentition, with significant psychosocial consequences *(Shipster et al., 2006 and Shipster et al., 2011 accepted for publication).*

The psychosocial and functional effects of macroglossia in BWS have been documented in Shipster et al. 2006 & Shipster et al. 2011 (accepted for publication). These two papers review the existing papers on functional and cosmetic outcomes in BWSm both pre and post surgery

Differing surgical techniques have been discussed by Davalbhakta and Lamberty 2000; Egyedi and Obwegeser 1964; Kacker et al 2000; Kveim et al 1985; Mixter et al 1993; Rizer et al 1985; Siddiqui and Pensler 1990; Tomlinson et al 2007.

2. Scope

2.1 Aims and objectives of service

The aim of this service is to provide a centralised, expert clinical service for children with macroglossia associated with Beckwith Wiedemann syndrome (BWSm).

This service is required for BWSm as there is currently no other service in the UK providing co-ordinated long-term care for these children. Our philosophy of care is to provide a seamless and comprehensive service and to ensure that the children and their families receive treatment that is seen to be the best current practice.

2.2 Service description/care pathway

The service includes four clinical specialities: speech and language therapy, surgery, orthodontics, and clinical nurse specialist.

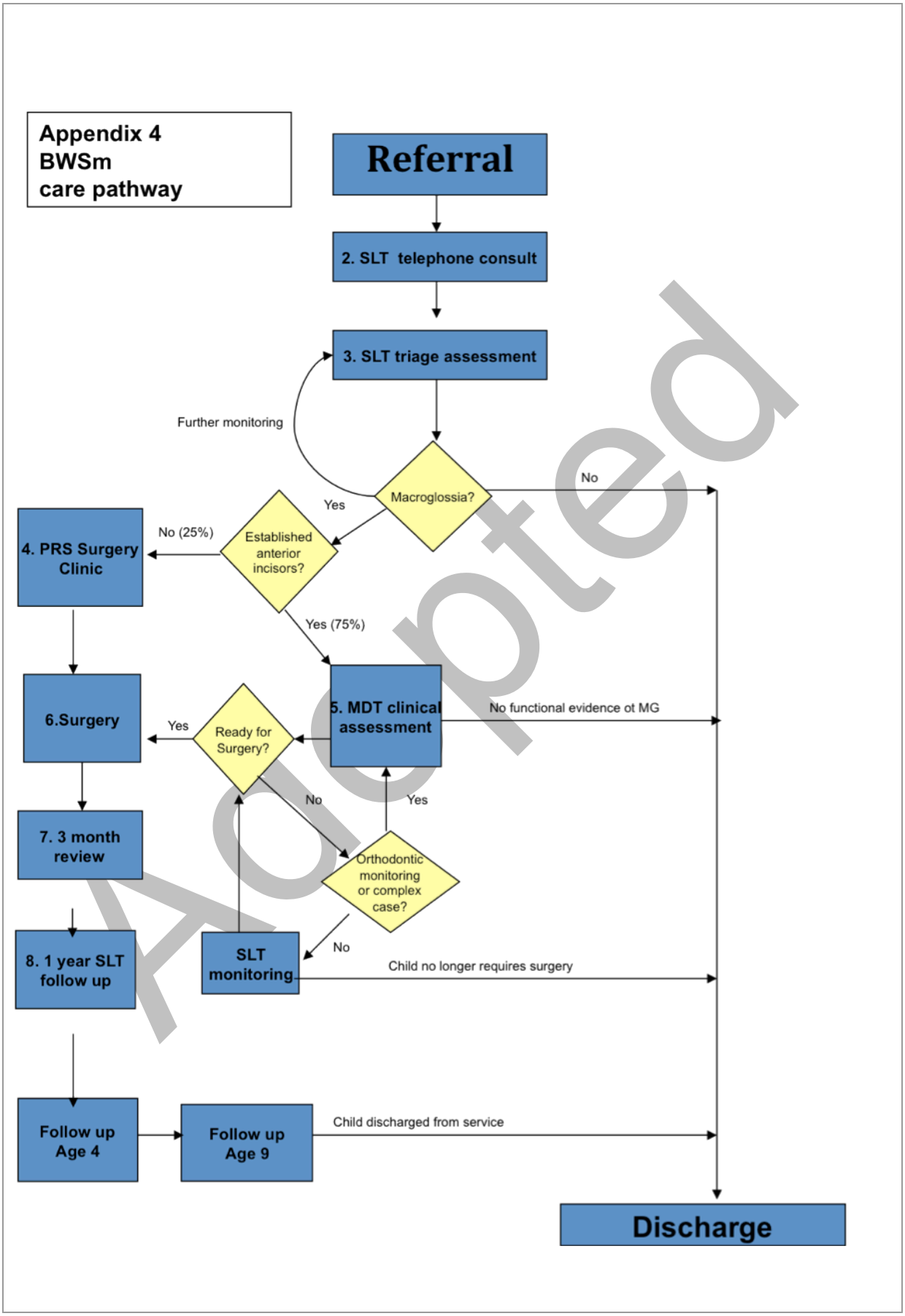
The service provides the following:

- assessment of the functional effects of macroglossia including: feeding, speech, drooling, oral motor skills, orthodontics and facial growth
- differential diagnosis between difficulties caused by the macroglossia and other developmental/medical problems
- specialist management advice, information on BWSm, the surgery procedure, the care pathway and contact details of other parents of children with BWSm who have a similar presentation to the child being assessed
- Speech and Language Therapy (SALT) acts as a triage and children are only put forward into the surgeon's clinic or multidisciplinary clinic at a point at which surgery can be considered. Some children will not require surgery and will be discharged from the service. Tongue reduction surgery is recommended for approximately 75% of the cases seen
- preparation for surgery in the pre-operative central nervous system (CNS) surgery clinic
- tongue reduction surgery
- feeding, speech, and wound care advice is provided during the post-operative recovery period
- advice and recommendations are given to local services regarding the

management of speech, feeding difficulties and orthodontic care. In many cases, unnecessary local intervention is avoided. For those children who do require speech and language therapy intervention, recommendations are made based on specialist knowledge of the condition and long-term outcomes and are therefore specific and time efficient for patients and local professions.

Enlargement of the tongue results from hyperplasia of muscle fibres, and whilst the tongue is generally increased in all three dimensions (Sokoloski et al.,1978; Vogel et al., 1986), there may also be individual variation in the presentation of the tongue (Shipster et al., 2006). BWSm is uniquely different from other forms of macroglossia (such as vascular anomalies) because the tongue tissue is normal.

To date, there are no objective techniques routinely used in the clinical diagnosis of macroglossia and the diagnosis is based on subjective criteria including the degree of tongue protrusion, and the clinical signs and symptoms that occur secondary to the macroglossia which are given in the table below. (Vogel et al., 1986; Wolford and Cottrell, 1996).



Functional signs and symptoms of macroglossia

1.1 Clinical areas	1.2 Characteristics associated with macroglossia
Respiratory/upper airway	airway obstruction, noisy breathing, dry cracked tongue
Orthodontic	anterior open bite, mandibular prognathism, class 111 malocclusion
Dental	widened interdental spaces, proclined anterior teeth, buccal tipping of posterior teeth
Drooling	inability to control oral secretions
Feeding	chewing and swallowing difficulties
Speech	articulation difficulties
Cosmetic/psychosocial	unusual facial appearance, child perceived as having learning difficulties
Other	scalloping of tongue edges, ulceration/secondary infection/haemorrhage of tongue

There is great variability in the presentation of the tongue in BWSm (Shipster et al., 2006).

For example, some tongues grow rapidly in the first year whereas others become satisfactorily accommodated in the oral structure. This variability is likely to be related to the various genotypes present in the condition (KvDMR hypomethylation, H19 hypermethylation, UPD 11p15, CDKNIC). However, due to the small numbers with a confirmed genetic diagnosis, the correlation between genotype and phenotype is not established.

At each assessment point, reports are written for professionals and parents summarising clinical findings and providing recommendations and management strategies where appropriate.

2.3 Population covered

The service outlined in this specification is for patients ordinarily resident in England, or otherwise the commissioning responsibility of the NHS in England (as defined in *Who Pays?:Establishing the responsible commissioner* and *other* Department of Health guidance relating to patients entitled to NHS care or exempt from charges).

NOTE:
for the purposes of commissioning health services this EXCLUDES patients who, whilst resident in England, are registered with a GP practice in Wales but includes patients resident in Wales who are registered with a GP practice in England.

2.4 Any acceptance and exclusion criteria

The child must have a diagnosis of macroglossia associated with Beckwith Wiedemann syndrome (BWS). Written clinical/genetic confirmation of BWS is required. A molecular diagnosis is currently present in 80% of cases (Elliot et al., 1994, Weksberg et al., 2005). The remaining 20% have the classic features of the condition but the cause is unknown.

The Child must be 6 months+ so that the macroglossia is stable. The macroglossia may become less evident in the first few months of life and in other cases; the tongue grows rapidly in the first few months of life.

The service appropriately manages patients with a physical or learning disability through reasonable adjustments and consideration of the holistic impact of treatment. This is taken into account during decision-making and the multi-disciplinary meeting.

For patients for whom English is not a first language, an interpreter service must be available.

Exclusion criteria:

- if the diagnosis is not Beckwith Wiedemann syndrome
- if macroglossia is not present.

Children are discharged from the service if they do not have functional evidence of macroglossia and if they have a combination of good speech, oral motor and dental outcomes.

All children are discharged from the service at nine years of age when secondary dentition is established and referred to local speech and orthodontic services if they have on-going problems in these areas.

2.5 Interdependencies with other services

The service is linked to charitable and patient-led organisations:
BWS Support Group.org.uk
Changing Faces: http//www.changingfaces.org.uk

3. Applicable Service Standards

3.1 Applicable national standards e.g. NICE, Royal College

The providers of the national BWS service must ensure they are fully integrated into their trust's corporate and clinical governance arrangements and must comply fully with Clinical Negligence Scheme for Trusts (CNST) and Care Quality Commission (CQC) requirements in terms of quality and governance. The hub centres are responsible for overseeing the governance arrangement of any spoke clinic provided under sub-contractual arrangements.

Each centre will ensure that there are:

- regular meetings with patient representatives;
- all practitioners will participate in continuous professional development and networking;
- patient outcome data is recorded and audited across the service.

4. Key Service Outcomes

Quality Performance Indicator	**Threshold**	**Method of measurement**	**Consequence of breach**	**Report Due**
Clinical Outcomes	To be agreed and developed in -year			
Speech	To be agreed and developed in -year			
Patient reported outcome measure (PROM)	To be agreed and developed in -year			

5. Location of Provider Premises

Great Ormond Street Hospital for Children NHS Foundation Trust