Scottish Paediatric Endocrine Group (SPEG)

Thyroid Dysfunction in patients with Down's Syndrome guideline

NOTE
This guideline is not intended to be construed or to serve as a standard of care. Standards of care are determined based on all clinical data available for an individual case and are subject to change as scientific knowledge and technology advance and patterns of care evolve. Adherence to guideline recommendations will not ensure a successful outcome in every case, nor should they be construed as including all proper methods of care or excluding other acceptable methods of care aimed at the same results. The ultimate judgement must be made by the appropriate healthcare professional(s) responsible for clinical decisions regarding a particular clinical procedure or treatment plan. This judgement should only be arrived at following discussion of the options with the patient, covering the diagnostic and treatment choices available. It is advised, however, that significant departures from the national guideline or any local guidelines derived from it should be fully documented in the patient's case notes at the time the relevant decision is taken.

Purpose of Guideline

- To guide the management of children with Down's Syndrome referred with an elevated (thyroid stimulating hormone) TSH concentration on Guthrie blood spot measurements
- to give guidance for ongoing management and monitoring of Down's infants and children with hypothyroidism
- if hyperthyroidism is suspected / confirmed, please contact your local paediatric endocrinologist

Who should use this document?

- Community Paediatricians, General paediatricians, paediatricians with an interest in paediatric endocrinology, paediatric endocrinologists
- this guideline is not designed for primary care use

Patients to whom this document applies

- children with Down's in Scotland who undergo TSH monitoring using blood spot measurements
- children with Down's in Scotland with an elevated TSH on blood spot measurements

Summary

Diagnosis

- children with Down's Syndrome are at increased risk of developing autoimmune conditions, including both hyper and hypothyroidism
- children with Down's Syndrome should have annual TSH measurements
- this can be performed by blood spot measurements
- if a raised TSH is found on blood spot measurements
    - additional investigations and assessments are required.

Confirmatory tests

- thyroid function tests (TSH and free levothyroxine – fT4, T3 /fT3)
- TPO (and where appropriate TSH receptor) antibodies*
    - * TSH receptor antibodies should only be requested if diagnostic query is hyperthyroidism

(For management of hypothyroidism – please see hypothyroidism guideline).

Assessment of Symptoms

These are non-specific and can be due to other factors.
They include:

- lethargy
- poor growth
- constipation

- weight gain
- behavioural change

The absence of clinical features does not exclude thyroid dysfunction.

The newborn screening laboratory will notify the designated person if the blood spot TSH is >6mu/l, or >4mu/l in children aged 1-5 years.

Depending on the result of the thyroid function tests and the presence/absence of symptoms will determine whether initiation of levothyroxine treatment is required (see flow diagram below)

LEVOTHYROXINE TREATMENT SHOULD BE STARTED

- if TSH >21mul/l
- if TSH >6 mu/l and the free T4 is <9pmol/l OR there are symptoms

LEVOTHYROXINE TREATMENT SHOULD BE ADVISED/CONSIDERED

- if TSH 11-20mu/l, but free T4 within normal limits and no symptoms
- if antibodies are present, treatment may also be considered
- if treatment not initiated, repeat TFTs in 6 months

LEVOTHYROXINE TREATMENT MAY NOT BE REQUIRED

- if TSH < 11mu/l with free T4 within normal limits and no symptoms

The above scenarios are a guide to when treatment with levothyroxine should be considered. If there are no clinical symptoms and the patient is well, they can be monitored every 6 months even in the presence of antibodies, as it may be some time before they develop hypothyroidism.

STARTING LEVOTHYROXINE TREATMENT

- levothyroxine requirement is approximately 100micrograms /m^2
- a starting dose of 25-50 micrograms usually given and titrated according to TFTs

Preparations of levothyroxine

Written instructions on how to give levothyroxine should be available and supplied to the parents.

Tablets

Available as 12.5 micrograms, 25 micrograms, 50 micrograms or 100 micrograms.
If unable to swallow tablets, they should be crushed and mixed with a small volume (≤ 5ml) of liquid or yoghurt.
Supply a tablet cutter for halving tablets if necessary.
The first dose should be given by parents under the supervision of a nurse or pharmacist.

Liquid preparations of levothyroxine are available however we recommend using levothyroxine in tablet form.

Tablets have been used extensively and successfully in the management of congenital hypothyroidism and therefore SPEG recommends the use of tablets, in accordance with ESPE guidelines [1]

CONTACT LIST

***First contact may not be a clinician and could be admin staff**

Centre	1st Contact*	Community Paediatrician	Paediatric Endocrinologist (switchboard)
Grampian	Claire Page 3rd Floor Royal Aberdeen Children's Hospital Westburn Road Aberdeen AB25 2ZG 01224 551 703	Dr Sue Moore 01224 710775	Amalia Mayo 0845 456 6000
Ayrshire	Amanda Brown 0129 432 3441	Community Paediatricians Rainbow House Ayrshire Central Hospital Irvine KA12 8SS	Scott Williamson 01563 521133

Centre	1st Contact*	Community Paediatrician	Paediatric Endocrinologist (switchboard)
Borders	Karen Purves Phone: 01896 826892 Or email: paediatric.secretaries@borders.scot.nhs.uk	Dr Graeme Eunson 01896 826 000	Dr Graeme Eunson 01896 826 000 Dr Andy Duncan 01896 826000
Dumfries	Dr Raj Shyam 01387 241732	Hospital Switchboard 01387 246246 and ask for on-call Consultant Paediatrician	Hospital Switchboard 01387 246246 and ask for on-call Consultant Paediatrician
Tayside	All negative reports should be sent to: Duty Team Blood Sciences NHS Tayside Ninewells Hospital Dundee DD1 9SY And not to Child Health	**Tayside Dundee** Dr Katherine Lawlor 0138 283 5100 **Tayside Angus** Dr Jenny Fraser 01307 47 5261	Nicky Conway 01382 660111
Lothian	Jackie Caldwell Community Child Health Dept. St John's Hospital, Howden Road West, Livingston, EH54 6PP 01506 524406	Endocrine admin team 0131 312 0443 who will link with Endocrine Consultant on duty. Or email rhcyp.endocrine@nhslothian.scot.nhs.uk	Endocrine admin team 0131 312 0443 who will link with Endocrine Consultant on duty. Or email rhcyp.endocrine@nhslothian.scot.nhs.uk
West Lothian	Jackie Caldwell Community Child Health Dept. St John's Hospital, Howden Road West, Livingston, EH54 6PP 01506 524406	Dr Jill Yates Community Child Health Dept. St John's Hospital, Howden Road West, Livingston, EH54 6PP 01506 524406	Endocrine admin team 0131 312 0443 who will link with Endocrine Consultant on duty. Or email rhcyp.endocrine@nhslothian.scot.nhs.uk

Centre	1st Contact*	Community Paediatrician	Paediatric Endocrinologist (switchboard)
East Lothian	Jackie Caldwell Community Child Health Dept. St John's Hospital, Howden Road West, Livingston, EH54 6PP 01506 524406	**East Lothian** Dr Jen McGill Community Child Health Department Musselburgh Primary Care Centre Inveresk Road Musselburgh, EH21 7BP 0131 446 4123	Endocrine admin team 0131 312 0443 who will link with Endocrine Consultant on duty. Or email rhcyp.endocrine@nhslothian.scot.nhs.uk
Midlothian	Jackie Caldwell Community Child Health Dept. St John's Hospital, Howden Road West, Livingston, EH54 6PP 01506 524406	**Midlothian** Dr Rachel Noble Community Child Health Musselburgh Primary Care Centre Inveresk Road Musselburgh EH21 7BP 0131 446 4131	Endocrine admin team 0131 312 0443 who will link with Endocrine Consultant on duty. Or email rhcyp.endocrine@nhslothian.scot.nhs.uk
Fife	Dr Berni Quinn 01383 623623	Dr Berni Quinn Queen Margaret Hospital, Whitefield Road, Dunfermline 01383 623623	Anthony Tasker 01592 643355
Glasgow	Gillian Barr 0141 201 0933	Sherin Hamza 0141 2010914	Guftar Shaikh 0141 201 0000/ 0141 451 6548
Highland	Dr Sheila Watt 01463 701337	Dr Sheila Watt Community Paediatrician Morven House Raigmore Hospital Inverness IV2 3UJ 01463 701337	Stuart Henderson 01463 704000
Lanarkshire	Steven Strachan 01698 752980	Dr Shewale 0169 836 6481	Dr Ian Hunter/ Dr Shubhangi Shewale 01698 8366481
Paisley	Kirsty Heron or Claire McEwan 0141 314 4662	Dr Lesley McDonald Panda Centre Aranthrue Centre, 103 Paisley Road, Renfrew PA4 8LH 0141 314 4603	Guftar Shaikh 0141 201 0000/ 0141 451 6548

Centre	1st Contact*	Community Paediatrician	Paediatric Endocrinologist (switchboard)
Argyll & Bute	Dr Caroline Clark Child Health Cowal Community Hospital 360 Argyll St Dunoon PA23 7RL	Dr Caroline Clark 01369 708346	Guftar Shaikh 0141 201 0000 / 0141 451 6548
Inverclyde	Debbie McLelland 01475 505140	Dr Alison Kelly 01475 505140	Guftar Shaikh 0141 201 0000/ 0141 451 6548
Forth Valley	Dr Mohamed Mansor 01324 567140	Dr Mohamed Mansor 01324 567140	Sabine Grosser 01324 567202 (bleep 1812)

SPEG - Management of Raised TSH Screening Result in Children With Downs Process (NSD610-016.14)