Williams Syndrome Clinical Management Guidelines

Management of Williams Syndrome

A Clinical Guideline

Williams Syndrome Guideline Development Group

williams SYNDROME FOUNDATION
www.williams-syndrome.org.uk

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Contents

Introduction…

… to Williams Syndrome (WS)

Williams Syndrome is a rare condition, that occurs in 1 in 20,000 births. The current definition of WS was agreed by the Williams Syndrome Guideline Development Committee at the Williams Syndrome Management Consensus Meeting held in Manchester in May 2009;

"Williams syndrome is a sporadic genetic disorder due to deletion of a small part of chromosome 7. Features may include a distinctive facial appearance, congenital heart defects and high levels of calcium in infancy. Early feeding problems are common and development is delayed. People with WS have sociable personalities, characteristic behavioural traits and variable degrees of learning disability."

… to the Williams Syndrome Guideline Development Project

The guidelines have been developed using a robust methodology based on the one utilised by the Scottish Intercollegiate Guidelines Network (SIGN). The method has been adapted to suit rare conditions where the evidence base is limited, and where expert consensus plays a greater role. The members of the guideline development group are listed on page 40.

… to the Williams Syndrome Clinical Management Guidelines

What are the aims of the guidelines?

The existing guidelines for the medical supervision of people with WS were published in 2001 by the American Academy of Pediatrics Committee on Genetics (AAPCG). Whilst a valued addition to the available guidance these are not entirely transferable to the UK. Therefore, in order to optimise the medical management of people with WS, the aim of the guidelines is to provide clear evidence-based management recommendations applicable to UK patients.

Who are they aimed at?

As WS is so rare, it is unlikely that the primary care clinicians usually responsible for coordinating the care of people with the condition will have had much prior experience of the syndrome. As it is a multisystem disorder, people with WS require various tests, screenings, assessments, referrals and multidisciplinary interventions at different stages of their lives. These guidelines lay out these requirements in a clear format that are accessible to anybody who is involved in the care of an individual with WS.

How are they used?

The guidelines are divided into recommendations for four age groups:
- Infancy: 0—1 year old
- Childhood: 1—11 years old
- Adolescence: 11—18 years old
- Adulthood: 18 years old +

On page 4, recommended baseline investigations are listed, to be consulted alongside the age group-specific recommendations at the time of diagnosis. For each age group, the recommended tests/screenings are listed, and follow-up options depending on the outcome of the test or screening are indicated.

NB. ABNL= Abnormal

Clinical features and recommended baseline investigations in Williams Syndrome

Clinical Features of Williams Syndrome	Baseline investigations (where investigation not indicated for a specific clinical feature, please refer to the relevant age group-specific page for management recommendations)
Confirm diagnosis of Williams Syndrome by testing for microdeletion on chromosome 7 using specialist molecular techniques e.g. FISH test*	
• Congenital heart defects (especially supravalvular aortic stenosis (SVAS) and peripheral pulmonary artery stenosis)	• Full cardiovascular assessment including scans and BP (blood pressure) measurement in both upper limbs.
• Raised blood/urine calcium levels	• Serum Ca and Urine Ca: Creatinine Ratio
• Nephrocalcinosis, bladder and renal tract abnormalities	• Renal tract ultrasound to include kidneys and bladder
• Endocrine abnormalities	• Thyroid Function Tests (TFTs)
• Failure to thrive/slow growth rate/feeding problems	• Plot growth on appropriate Williams Syndrome growth chart (see pages 14 and 15)
• Hypertension • Scoliosis and other musculoskeletal problems • Gastrointestinal problems • Distinctive facial features • Dental anomalies • Distinctive behavioural characteristics including irritability, anxiety, overfriendliness • Hypersensitivity to noise	

*Fluorescence in situ hybridisation (FISH) is the most common, but not the only available test for confirming a diagnosis of Williams Syndrome. Some laboratories may use other DNA-based diagnostic techniques.

Recommendations for the management of Williams Syndrome

~ in neonates & infancy (1) ~

AGE 0—1

Recommended Testing/Screening		Clinical Management Recommendations
• **Serum Ca and Urine Ca: creatinine ratio**	→	If normal and under 1 year old, repeat test at 12 months.
	ABNL →	Management of Hypercalcaemia - calcium intake should be equal to or less than half of the recommended nutrient intake (RNI) for the patient's age group. - stop use of supplements containing calcium. - Ensure that infant feeds are prepared using 'soft' water. - Ensure adequate rehydration. - Locasol formula milk (SHS Nutrition) - Steroids (Prenisolone), orally as necessary. - Monitor blood pressure - take sunblock if travelling/in sunny conditions. - 3 monthly follow up. - If serum PTH starts to rise, relax calcium intake but monitor blood and urine calcium levels. - Consider referral to paediatric metabolic bone disorder specialist. - In rare cases, where hypercalcaemia is refractory to hydration and low-calcium diet, intravenous Pamidronate may be necessary.
• **Thyroid Function Tests (TFTs)**	→	Ensure baseline test undertaken. Repeat thyroid function test if patient symptomatic.
	ABNL →	Measure TSH levels and if elevated, consider thyroid scanning.
• **Renal tract screening to include kidneys and bladder**	ABNL →	If nephrocalcinosis refer to nephrologist for 6 monthly screening. If structural abnormalities, management or referral as necessary.
• **Hypertension screening**	→	Annual monitoring of blood pressure in both upper limbs and left leg.
	ABNL →	If associated with renal artery stenosis (RAS), refer to nephrologist. Surgical treatment where necessary. **NB.** If RAS is present, angioplasty is not recommended due to elastinopathy. If essential hypertension, manage with calcium channel blockers where medical management is required (and RAS has been ruled out).
• **Cardiac screening**	→	Full cardiac assessment including scans before one year old if diagnosis made in neonatal period. Annual cardiac examination until 4 years old. Follow up by cardiologist.

Recommendations for the management of Williams Syndrome

~ in neonates & infancy (2) ~

AGE 0—1

Recommended Testing/Screening		Clinical Management Recommendations
• **Feeding & Gastrointestinal issues**	→	Take feeding history. Enquire about bowel habit.
	ABNL →	If problems, refer for appropriate support and treat constipation.
• **Growth**	→	Measure height, weight and occipitofrontal circumference (OFC) at birth and 1-3 monthly.
	ABNL →	Routine paediatric investigations for failure to thrive and reduced growth velocity.
• **Hearing screening**	→	NHS newborn hearing screening programmes throughout UK (NHSP) - screening within the first few weeks.
• **Vision screening**	→	Visual screening should take place between 6 and 12 months. Parents to report any concerns.
	ABNL →	Refer to community optometric/orthoptic service (via GP) if abnormality found.
• **Screening for dental anomalies**	→	Enrol patient in an individualised preventative oral healthcare programme from an early age. Routine follow up and regular dental examinations by a family dentist or local community dental services are essential.
	ABNL →	Missing teeth/malocclusion/other dental anomalies: refer to a consultant in paediatric dentistry for multidisciplinary management.
• **Multidisciplinary developmental assessment**	→	Between 0-3 years old. Coordinated by hospital or community paediatrician.

! Anaesthesia

A paediatric anaesthetist should be involved in the pre-op care of children up to 3-4 years old.
Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.
Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.

Recommendations for the management of Williams Syndrome

~ in childhood (1) ~

AGE 1—11

Recommended Testing/Screening		Clinical Management Recommendations
• **Serum creatinine**	→	In all WS children, test serum creatinine every 2—4 years.
	ABNL →	Investigate/refer as appropriate—check for infection, exclude obstructive lesion(s), undertake detailed renal function tests and/or refer to a nephrologist.
• **Serum Ca and Urine Ca: creatinine ratio**	→	If normal when under 1 year old, repeat test at 12 months.
	ABNL →	Management of Hypercalcaemia - Calcium intake should be equal to or less than half of the recommended nutrient intake (RNI) for the patient's age group. - Stop use of supplements containing calcium. - Ensure that infant feeds are prepared using 'soft' water. - Ensure adequate rehydration. - Locasol formula milk (SHS Nutrition) - Steroids (Prenisolone), orally as necessary. - Monitor blood pressure - Take sunblock if travelling/in sunny conditions. - 3 monthly follow up. - If serum PTH starts to rise, relax calcium intake but monitor blood and urine calcium levels. - Consider referral to paediatric metabolic bone disorder specialist. - In rare cases, where hypercalcaemia is refractory to hydration and low-calcium diet, intravenous Pamidronate may be necessary.
		Monitor for 1—2 years after hypercalcaemia has resolved.
• **Thyroid Function Tests (TFTs)**	→	Test if patient is symptomatic.
	ABNL →	Measure TSH levels and if elevated, consider thyroid scanning, Consider referral to endocrinologist for treatment with L-Thyroxine if patient has overt hypothyroidism, or progressive deterioration of thyroid function.
• **Renal screening**	→	Renal tract ultrasound to include kidneys and bladder if symptomatic.
	ABNL →	If nephrocalcinosis refer to nephrologist for 6 monthly screening.
• **Hypertension screening**	→	Annual monitoring of blood pressure in both upper limbs and left leg.
	ABNL →	If associated with renal artery stenosis (RAS), refer to nephrologist. Surgical treatment where necessary. **NB.** If RAS is present, angioplasty is not recommended due to elastinopathy. If essential hypertension, manage with calcium channel blockers where medical management is required (and RAS has been ruled out).

Recommendations for the management of Williams Syndrome

~ in childhood (2) ~

AGE 1—11

Recommended Testing/Screening		Clinical Management Recommendations
• **Cardiac screening**	→	Annual cardiac examination until 4 years old, and once between 5-13 years old. Full cardiac assessment including scans every 5 years.
• **Feeding & Gastrointestinal issues**	→	Enquire about feeding problems annually. Enquire about bowel habit annually.
	ABNL →	Treat constipation.
• **Screen for coeliac disease**	→	Once, after 3 years of age, with low threshold to repeat if suggestive symptoms.
• **Growth & Puberty**	→	Height, weight and OFC measurements 1-3 monthly until 2 years of age. Annually thereafter (use WS growth charts). Mid parental height centile should be estimated. Check spine clinically for kypho/scoliosis at puberty and x-ray/refer to orthopaedic team as indicated.
	ABNL →	Routine paediatric investigations for abnormal growth velocity and precocious puberty (< 8 years). Where necessary, consider gonadotropin releasing hormone (GnRH) therapy.
• **Hearing screening**	→	18 months: screen for otitis media with effusion (OME) & hyperacusis 3 years: screen for OME & language development 5-10 years: screen for hyperacusis & hearing loss 11-18 years: screen for hyperacusis & high frequency hearing loss
	ABNL →	If hyperacusis, implement a programme of desensitisation (plus maskers if necessary).
• **Vision screening**	→	Visual screening should take place between 6 and 12 months. Parents to report any concerns.
	ABNL →	Refer to community optometric/orthoptic service (via GP) if abnormality found.
• **Screening for dental anomalies**	→	Enrol patient in an individualised preventative oral healthcare programme from an early age. Routine follow up and regular dental examinations by a family dentist or local community dental services are essential.
	ABNL →	Missing teeth/malocclusion/other dental anomalies: refer to a consultant in paediatric dentistry for multidisciplinary management.

Recommendations for the management of Williams Syndrome

~ in childhood (3) ~

AGE 1—11

Recommended Testing/Screening	Clinical Management Recommendations
• **Multidisciplinary developmental assessment**	Between 0-3 years old. Should involve local Child Development/ Learning Difficulties (LD) Teams. Involve Child and Adolescent Mental Health Services (CAMHS) if necessary.
	Ongoing review and support of learning and development with further assessment of special educational needs as required.
• **Behavioural & Mental Health issues**	Behavioural management advice and support to family as required. Refer for psychological intervention for anxiety, and when major life events.

! Anaesthesia

A paediatric anaesthetist should be involved in the pre-op care of children up to 3-4 years old.
Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.
Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.

Recommendations for the management of Williams Syndrome

~ in adolescence (1) ~

AGE 11-18

Recommended Testing/Screening		Clinical Management Recommendations
• **Serum creatinine**		In all WS adolescents, test serum creatinine every 2—4 years.
	ABNL	Investigate/refer as appropriate—check for infection, exclude obstructive lesion(s), undertake detailed renal function tests and/or refer to a nephrologist.
• **Serum Ca and Urine Ca: creatinine ratio**		Test if symptomatic of hypercalcaemia.
	ABNL	If abnormal, investigate and manage as appropriate.
• **Thyroid Function Tests (TFTs)**		Test if patient is symptomatic.
	ABNL	Measure TSH levels and if elevated, consider thyroid scanning, Consider referral to endocrinologist for treatment with L-Thyroxine if patient has overt hypothyroidism, or progressive deterioration of thyroid function.
• **Renal screening**		Renal tract ultrasound at puberty, before leaving paediatric care, and 5 yearly thereafter, or if symptomatic.
	ABNL	If nephrocalcinosis refer to nephrologist for 6 monthly screening.
• **Hypertension screening**		Annual monitoring of blood pressure.
	ABNL	If associated with renal artery stenosis (RAS), refer to nephrologist. Surgical treatment where necessary. **NB.** If RAS is present, angioplasty is not recommended due to elastinopathy.
		If essential hypertension, manage with calcium channel blockers where medical management is required (and RAS has been ruled out).
• **Cardiac screening**		Cardiac examination once between 5-13 years old and 13-21 years old (follow up if symptomatic). Full cardiac assessment including scans every 5 years.
• **Gastrointestinal issues**		Enquire about bowel habit annually.
	ABNL	Treat constipation and consider investigating for diverticular disease.

Recommendations for the management of Williams Syndrome

~ in adolescence (2) ~

AGE 11-18

Recommended Testing/Screening		Clinical Management Recommendations
• **Screen for coeliac disease**	→	If patient symptomatic.
• **Growth & Sexual Health**	→	Check spine clinically for kypho/scoliosis at puberty and x-ray/refer to orthopaedic team as indicated. Weigh annually, and avoid excessive weight gain. Offer contraceptive advice/contact details of organisations who can advise on contraception for people with learning disabilities.
	ABNL →	Consider GnRH therapy for precocious puberty.
• **Hearing screening**	→	11-18 years: screen for hyperacusis & high frequency hearing loss
	ABNL →	If hyperacusis, implement a programme of desensitisation (plus maskers if necessary).
• **Screening for dental anomalies**	→	Ensure patient enrolled in an individualised preventative oral healthcare programme. Routine follow up and regular dental examinations by a family dentist or local community dental services are essential.
	ABNL →	Missing teeth/malocclusion/other dental anomalies: refer to a consultant in paediatric dentistry for multidisciplinary management.
• **Multidisciplinary developmental assessment**	→	Should involve local Child Development/ Learning Difficulties (LD) Teams. Involve Child and Adolescent Mental Health Services (CAMHS) if necessary. Ongoing review and support of learning and development with further assessment of special educational needs as required. Referral to local Connexions service may be appropriate/helpful.
• **Behavioural & Mental Health issues**	→	Behavioural management advice/support for family as required. Access to social skills training, and programmes to teach basic self help and daily living skills. Refer for psychological intervention for anxiety, and when major life events. **NB.** Apparent friendliness and sociability can mask depression and anxiety.

! Anaesthesia

Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.
Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.

Recommendations for the management of Williams Syndrome

~ in adulthood (1) ~

AGE 18+

Recommended Testing/Screening		Clinical Management Recommendations
• **Serum creatinine**		In all WS adults, test serum creatinine every 2—4 years.
	ABNL	Investigate/refer as appropriate—check for infection, exclude obstructive lesion(s), undertake detailed renal function tests and/or refer to a nephrologist.
• **Serum Ca and Urine Ca: creatinine ratio**		Test if symptomatic of hypercalcaemia.
	ABNL	If abnormal, investigate and manage as appropriate.
• **Thyroid Function Tests (TFTs)**		Test if/when patient is symptomatic, and check for anti-thyroid antibodies.
	ABNL	Measure TSH levels and if elevated, consider thyroid scanning, If compensated hypothyroidism present, refer to endocrinologist and monitor TFT and TSH annually. If TSH level significantly low, consider thyroid replacement therapy.
• **Renal screening**		Bladder & kidney ultrasonography every 5 years and if/when symptomatic.
	ABNL	If nephrocalcinosis refer to nephrologist for 6 monthly screening.
• **Hypertension screening**		Annual monitoring of blood pressure.
	ABNL	If associated with renal artery stenosis (RAS), refer to nephrologist. Surgical treatment where necessary. **NB.** If RAS is present, angioplasty is not recommended due to elastinopathy.
		If essential hypertension, manage with calcium channel blockers where medical management is required (and RAS has been ruled out). Consider referral to renal specialist for care of adults with hypertension.
• **Cardiac screening**		Full assessment including scans, every 5 years throughout life.

Recommendations for the management of Williams Syndrome

~ in adulthood (2) ~

AGE 18+

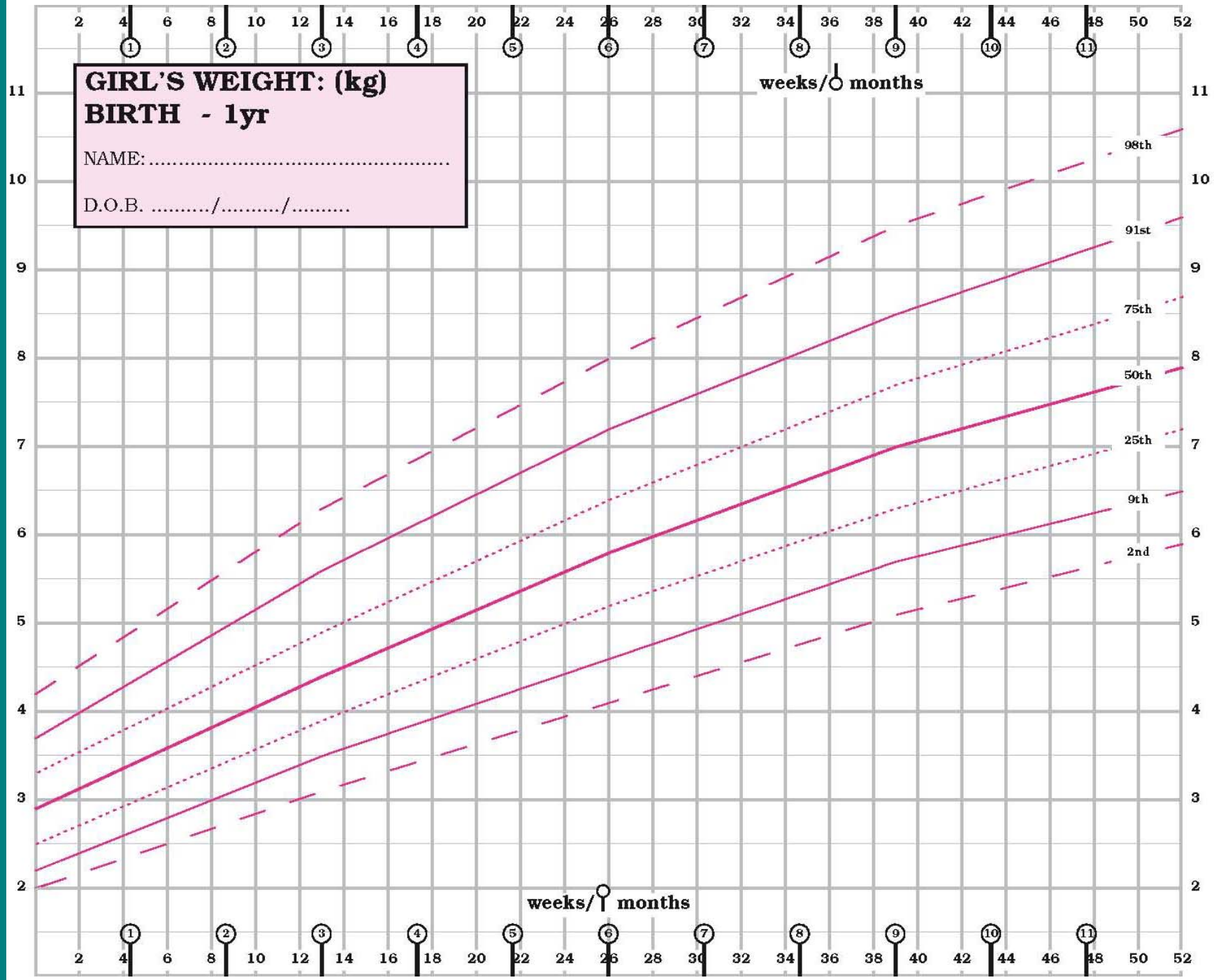
Recommended Testing/Screening		Clinical Management Recommendations
• **Gastrointestinal issues**		Enquire about bowel habit annually.
	ABNL	Treat constipation and consider investigating for diverticular disease.
• **Screen for coeliac disease**		If patient symptomatic.
• **Screening for diabetes**		At 30 years old: Oral Glucose Tolerance Test (OGTT), (or fasting insulin if considered more appropriate). Repeat OGTT if rapid weight gain. NB/ Do **not** use haemoglobin A1C as a screening tool.
	ABNL	Control impaired glucose tolerance with exercise & diet. Avoid large glucose loads over a short time period. Avoid diabetogenic drugs. Manage clinical diabetes in WS in the same way as in general population.
• **Growth & Sexual Health**		Weigh annually, and avoid excessive weight gain—encourage an 'active' lifestyle. Offer contraceptive advice/contact details of organisations who can advise on contraception for people with learning disabilities.
• **Hearing screening**		Every 10 years (for hearing loss and wax build-up).
• **Screening for dental anomalies**		Routine follow up and regular dental examinations by a family dentist or local community dental services are essential.
	ABNL	Missing teeth/malocclusion/other dental anomalies: refer to a consultant in Adult Restorative Dentistry or Special Care for multidisciplinary management.
• **Behavioural & Mental Health issues**		Access to support for employment, self help and independent living. Social skills intervention as needed. Refer for psychological intervention/support for anxiety, and when major life events. **NB.** Apparent friendliness and sociability can mask depression and anxiety.

! Anaesthesia

Unless there are existing cardiac problems, cardiac assessment within 12 months prior to a general anaesthetic is sufficient.
Pre-op assessment should take place 1-2 weeks prior to planned surgery, to assess cardiac, airway, joints, renal and emotional status.

Williams Syndrome Growth Charts

From: Martin, N. D. T., W. R. Smith, et al. (2007). *"New height, weight and head circumference charts for British children with Williams syndrome."* **Arch Dis Child 92(7): 598-601.**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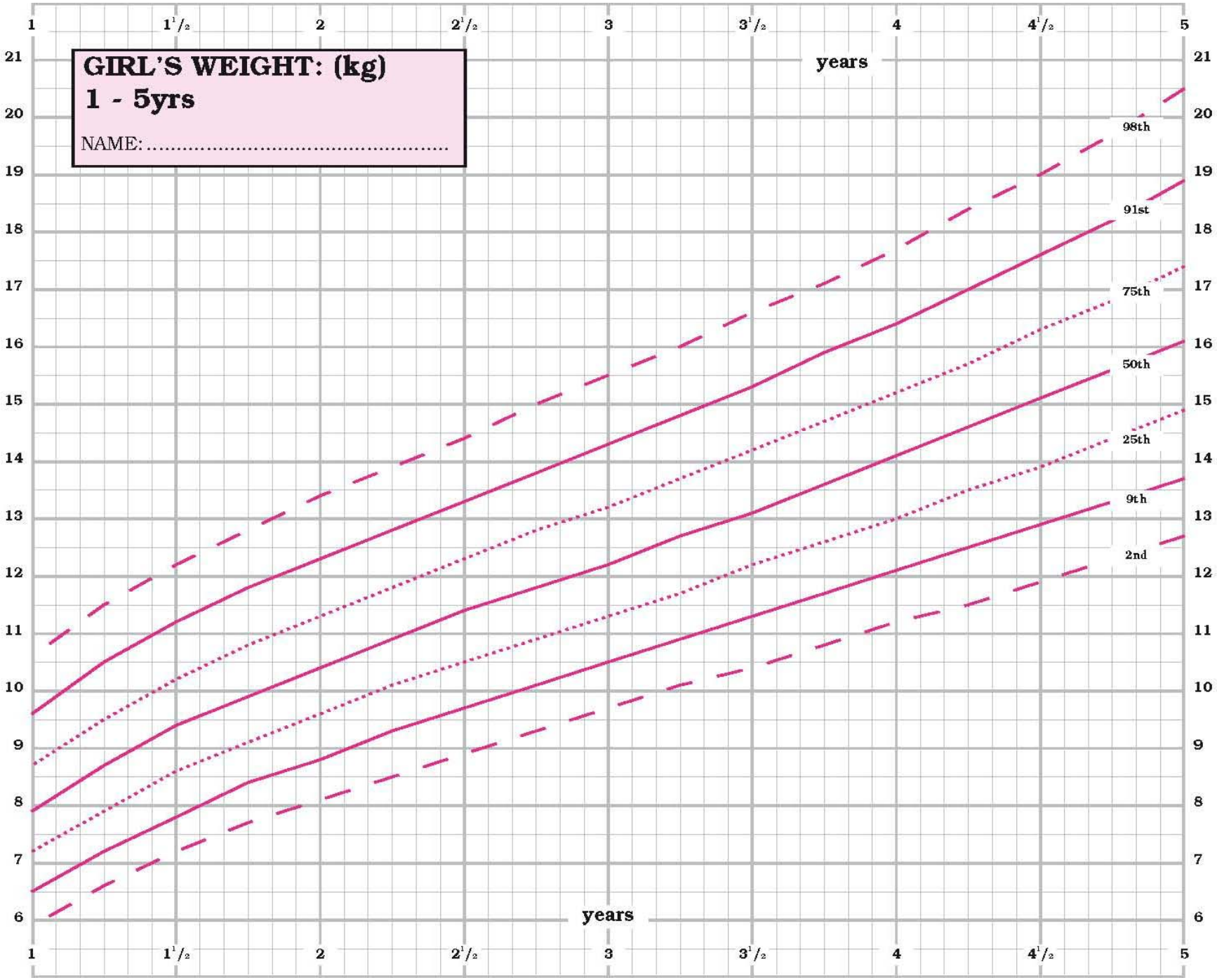
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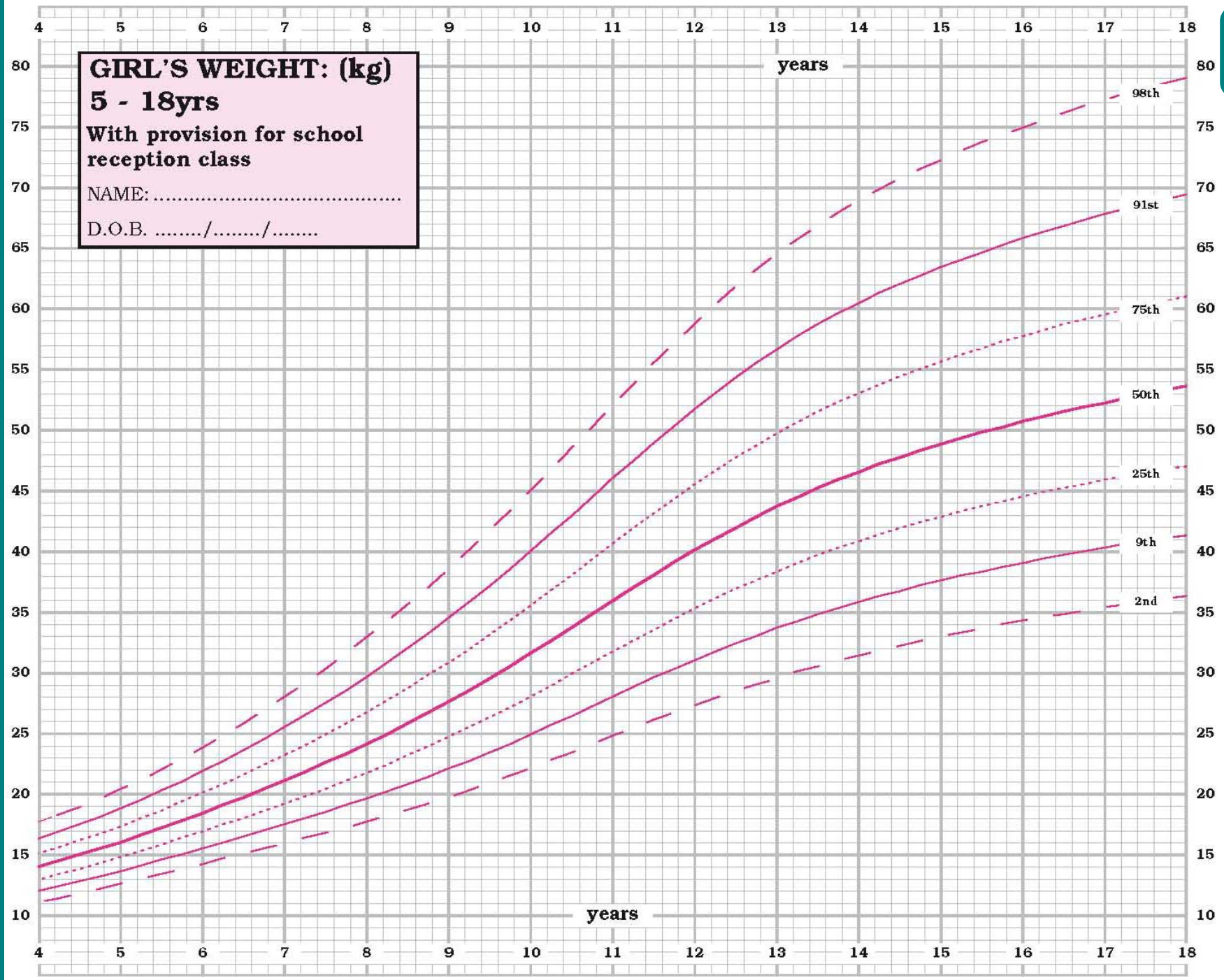
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5 - 18yrs
With provision for school reception class
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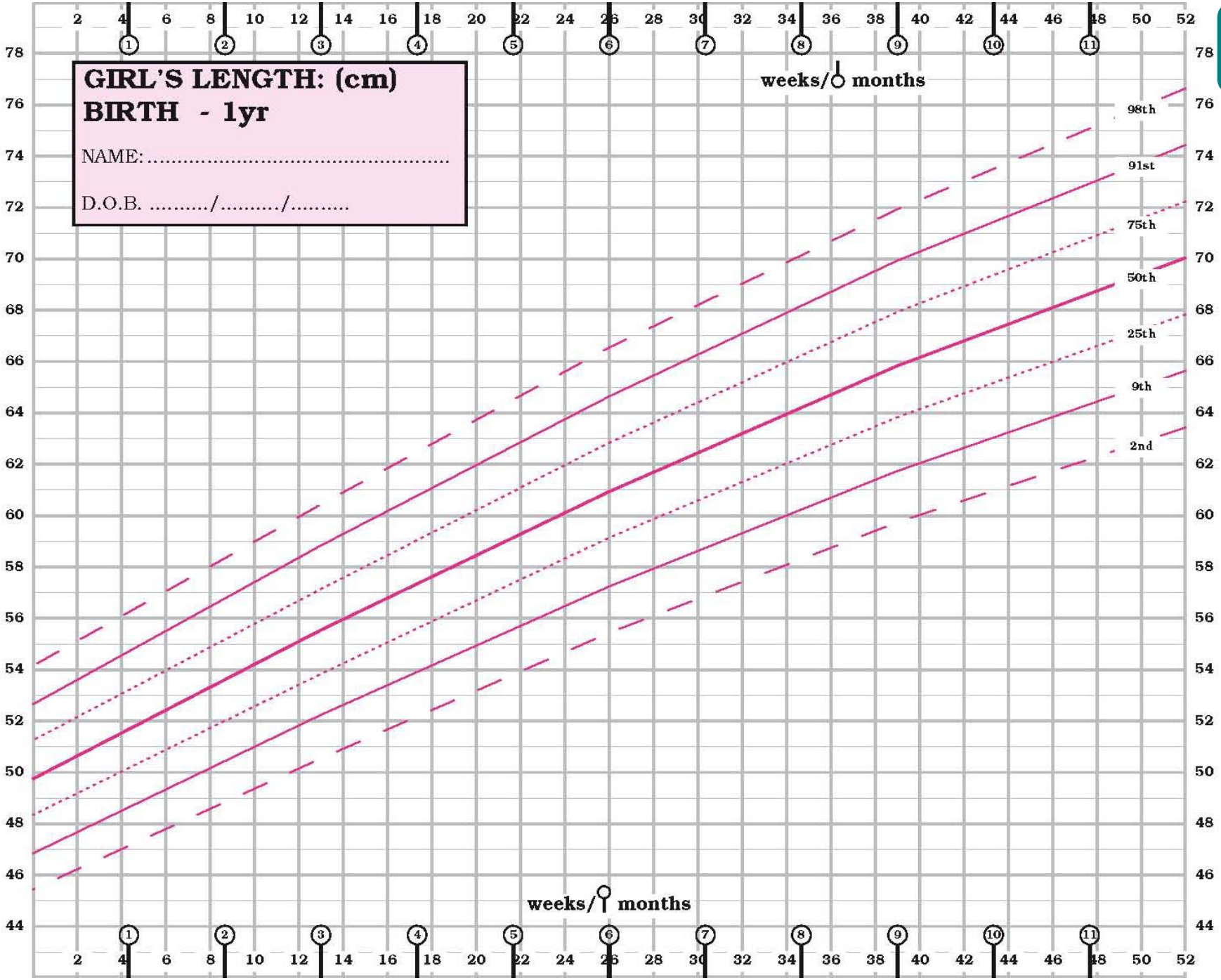
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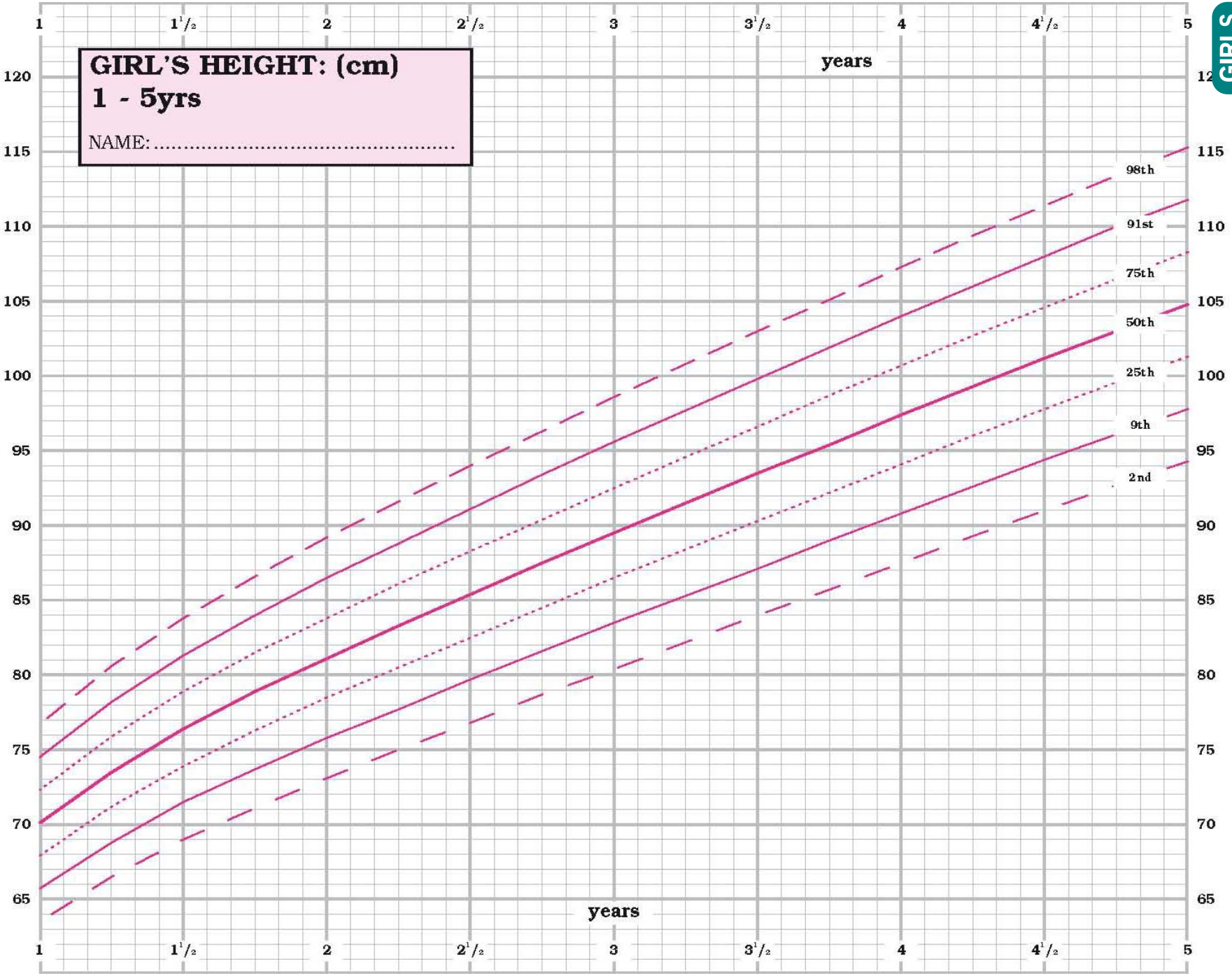
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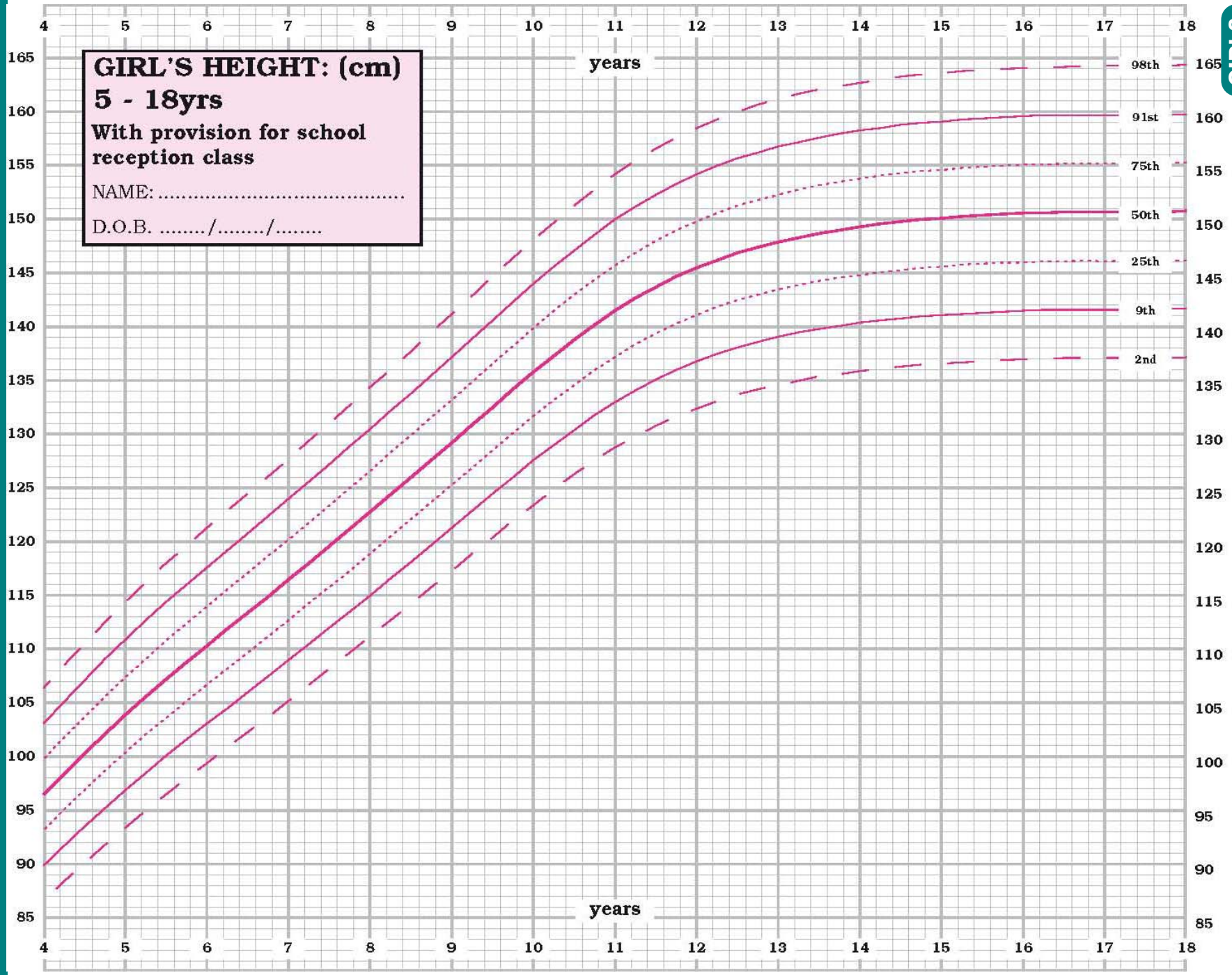
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With provision for school reception class
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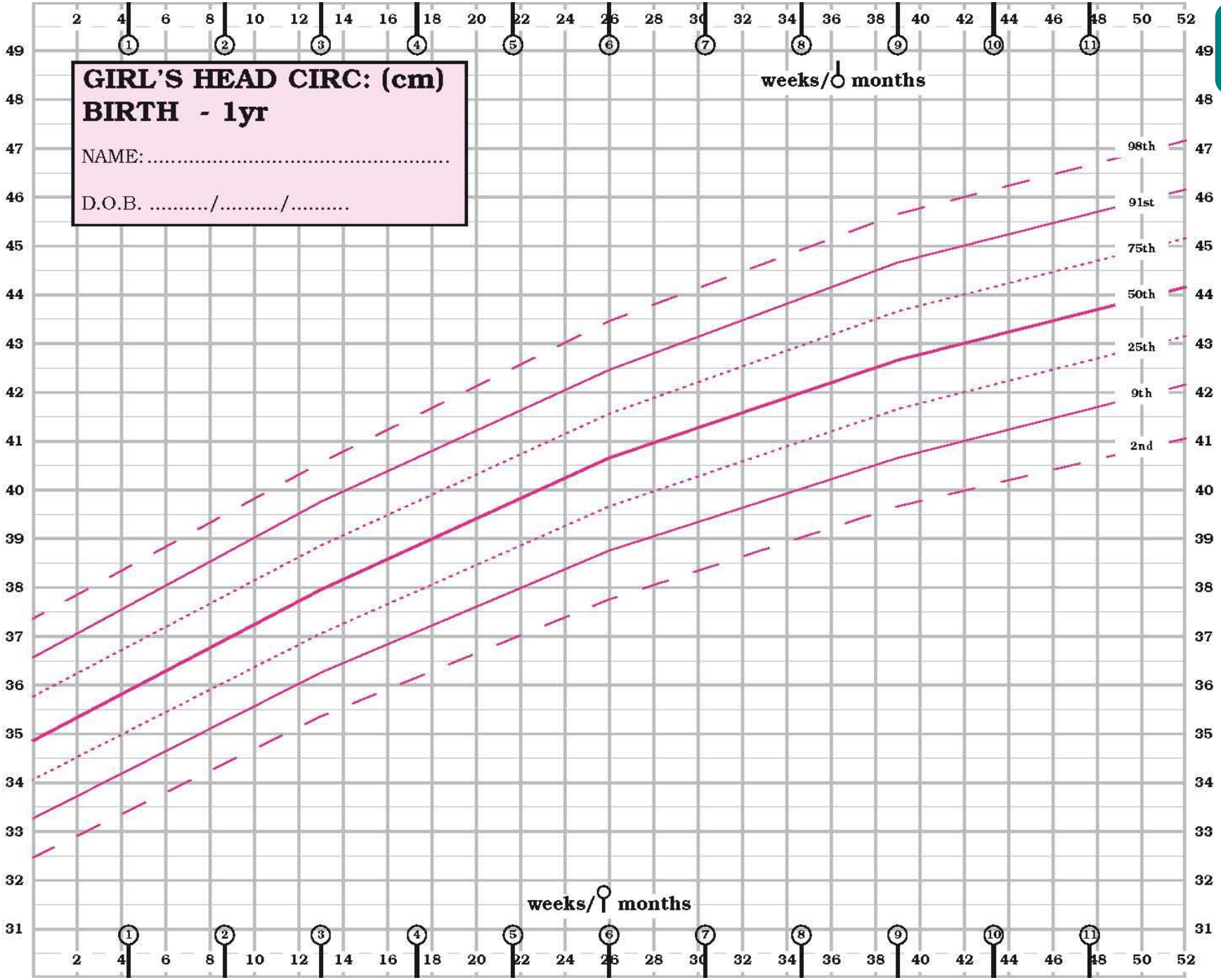
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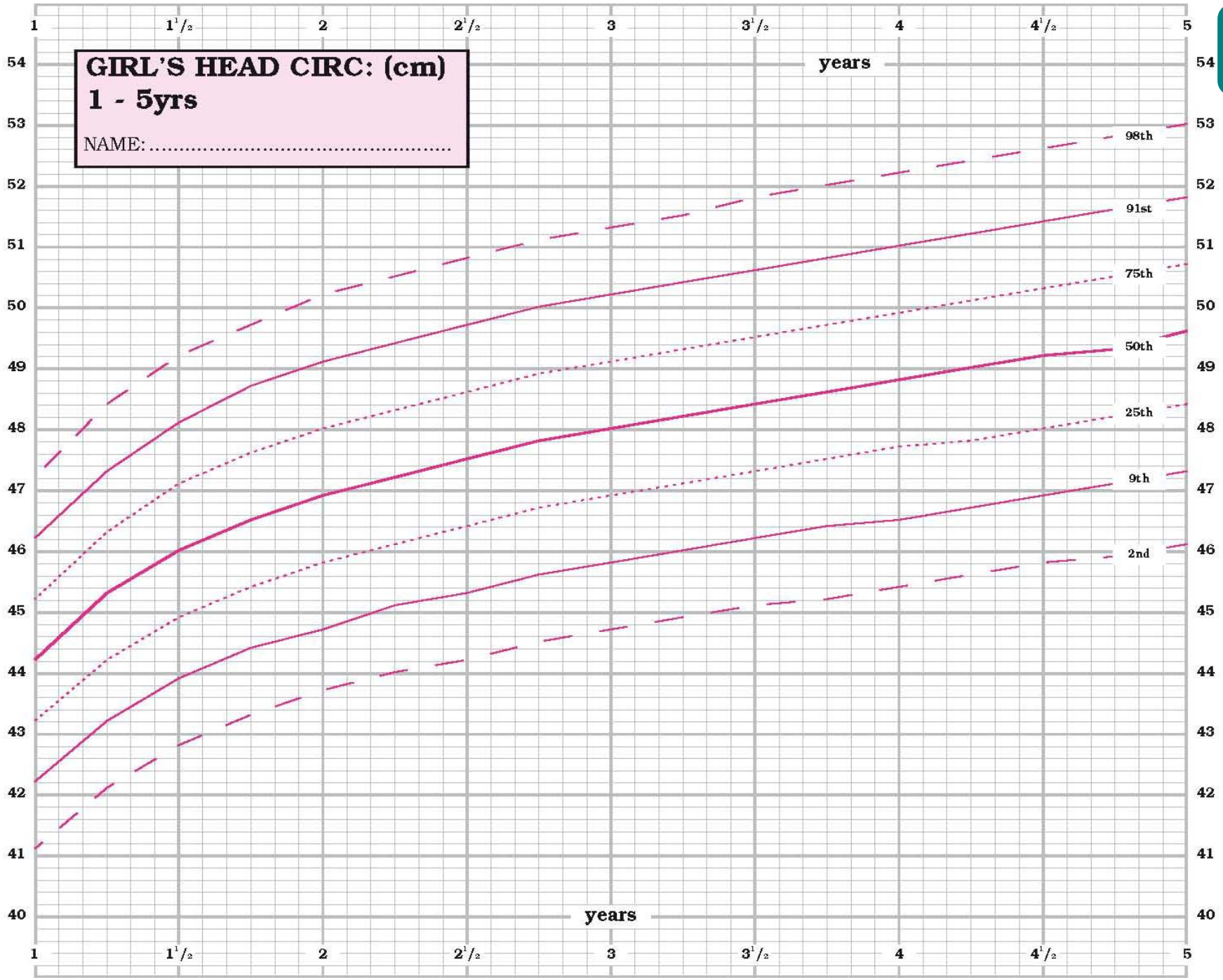
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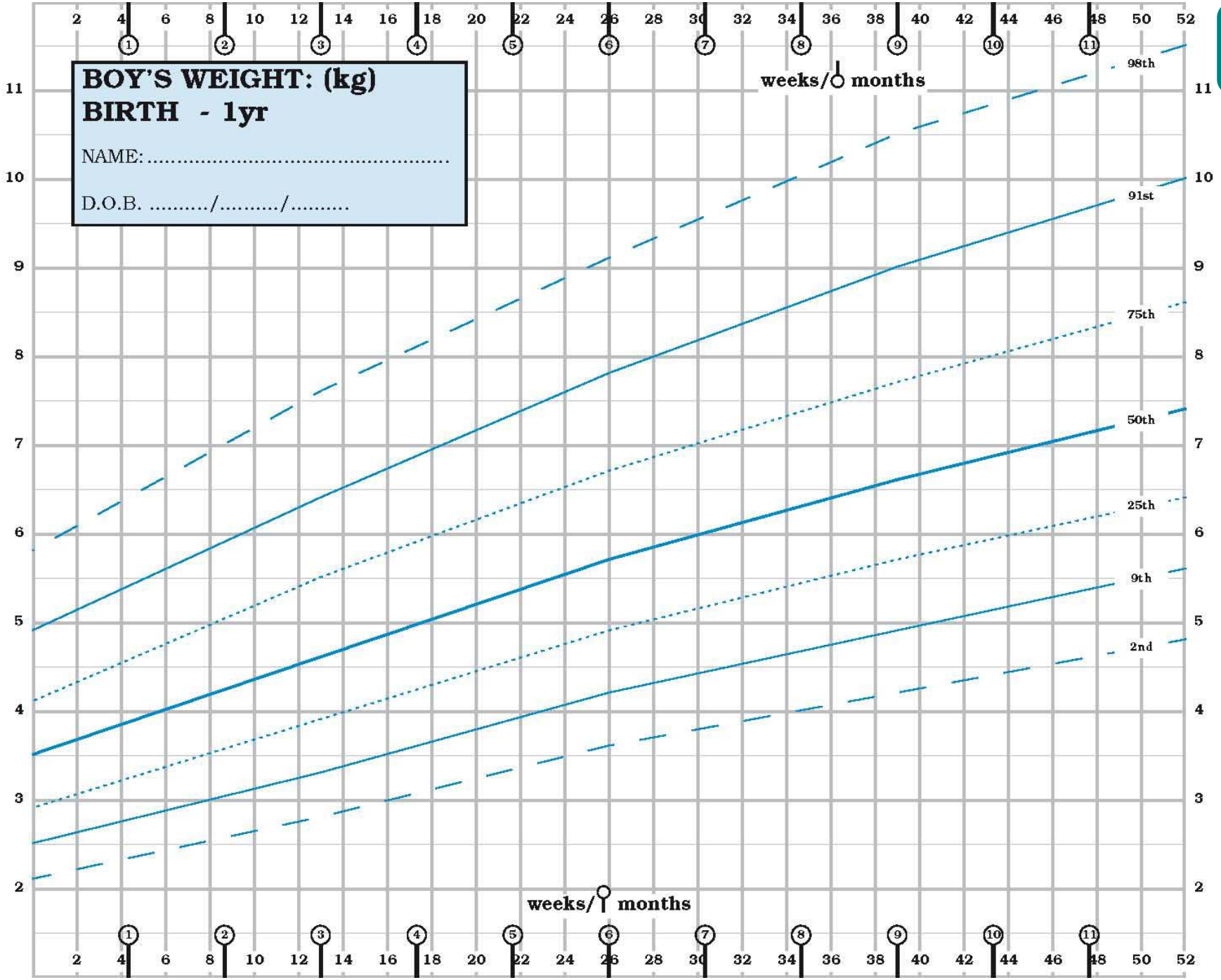
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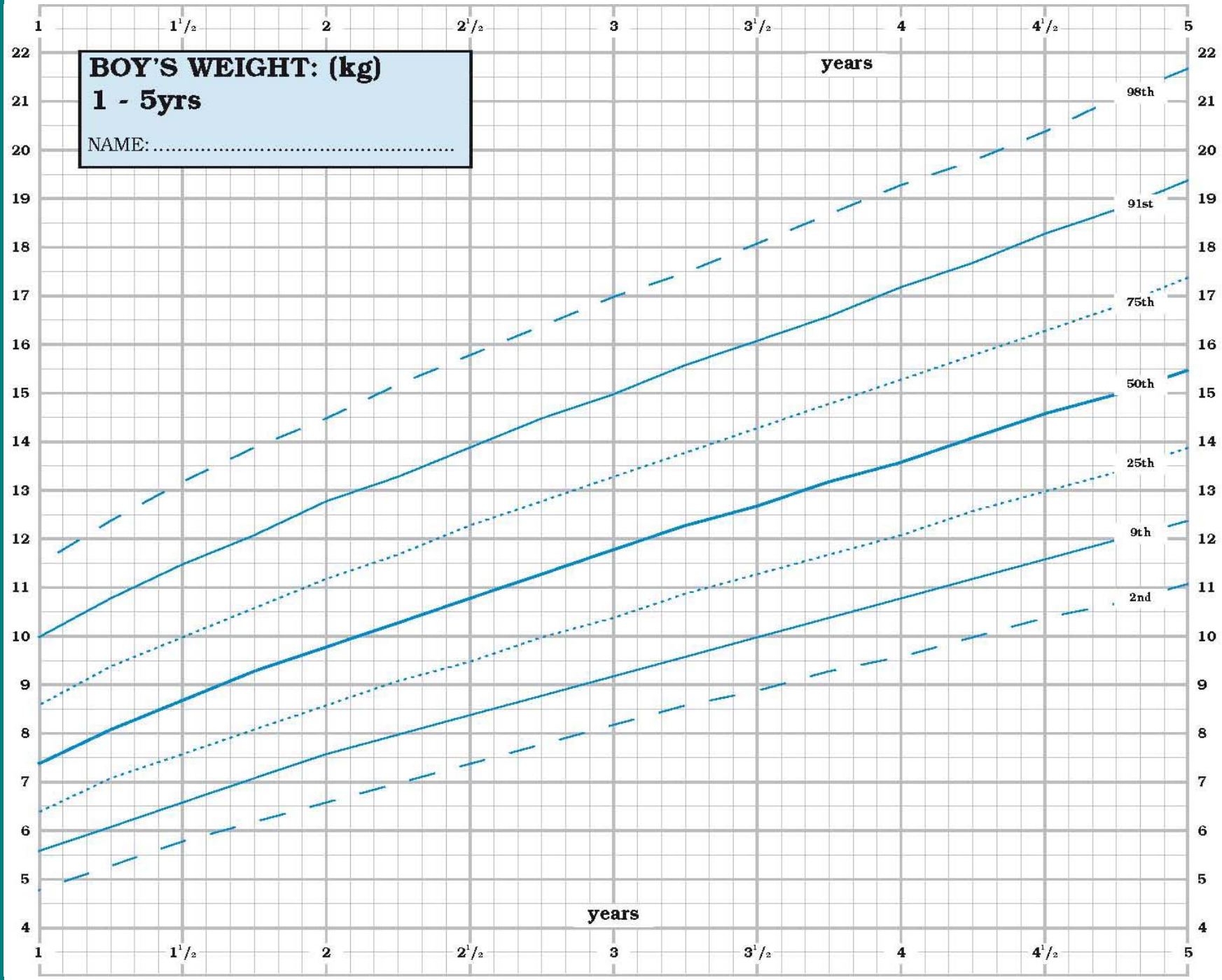
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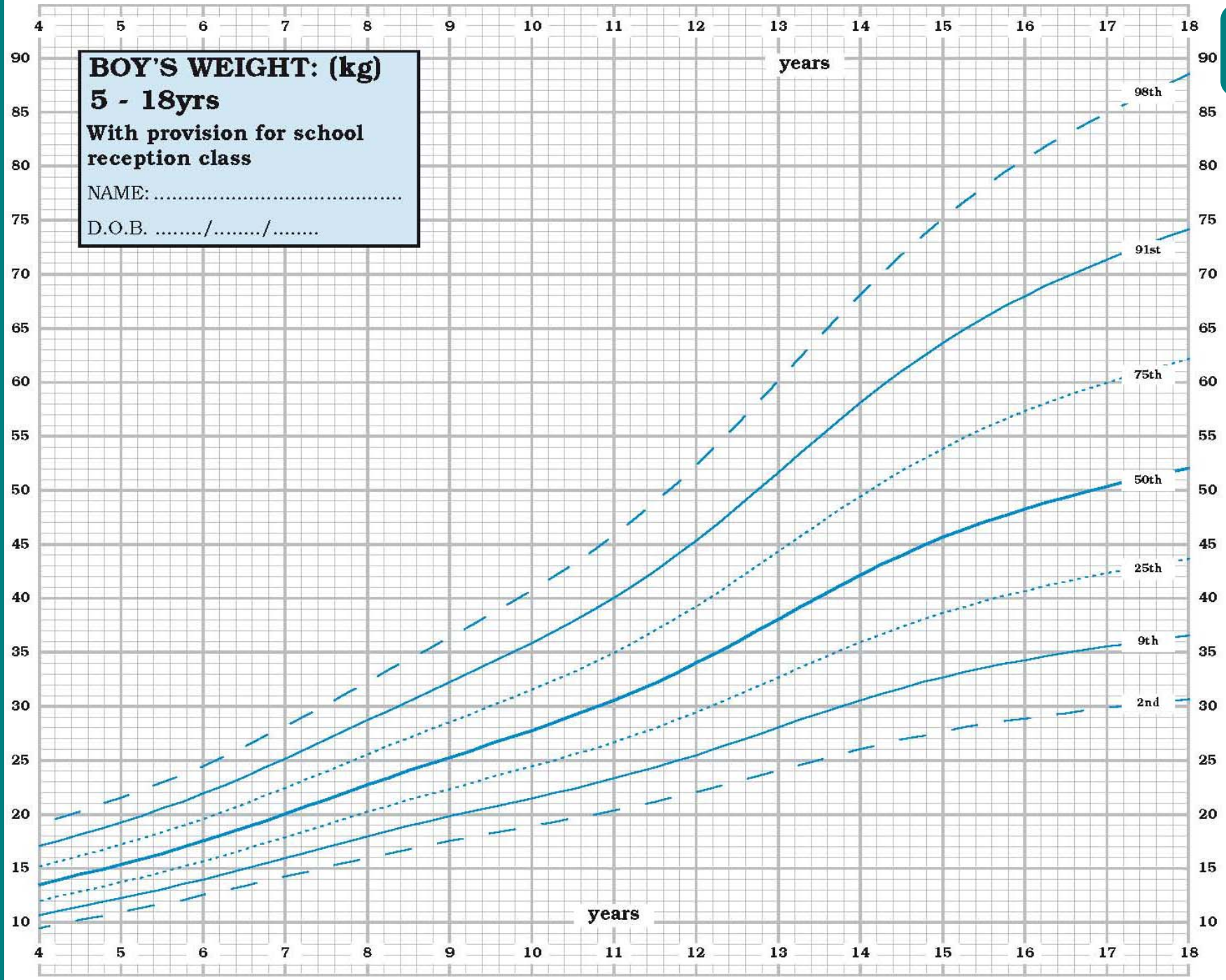
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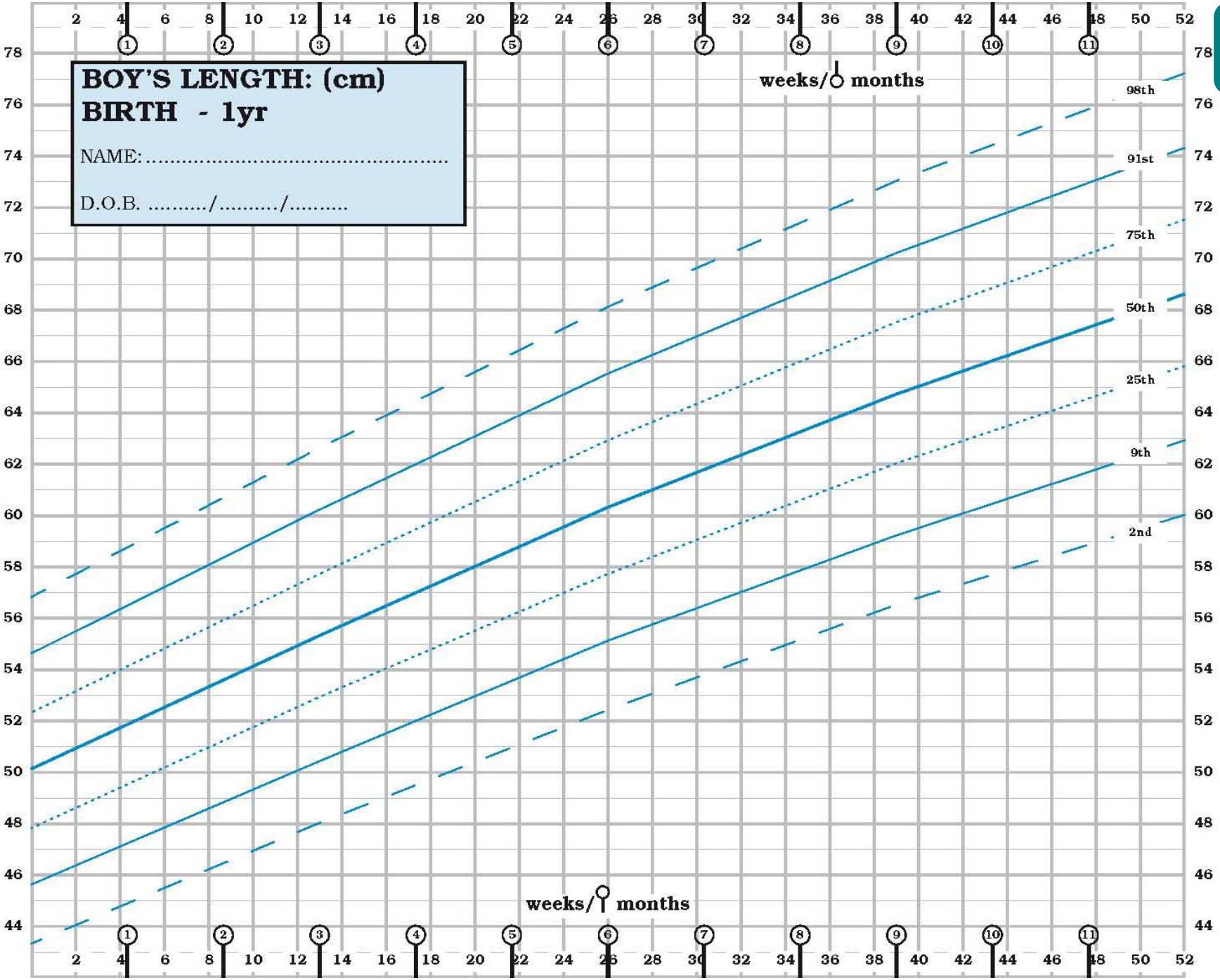
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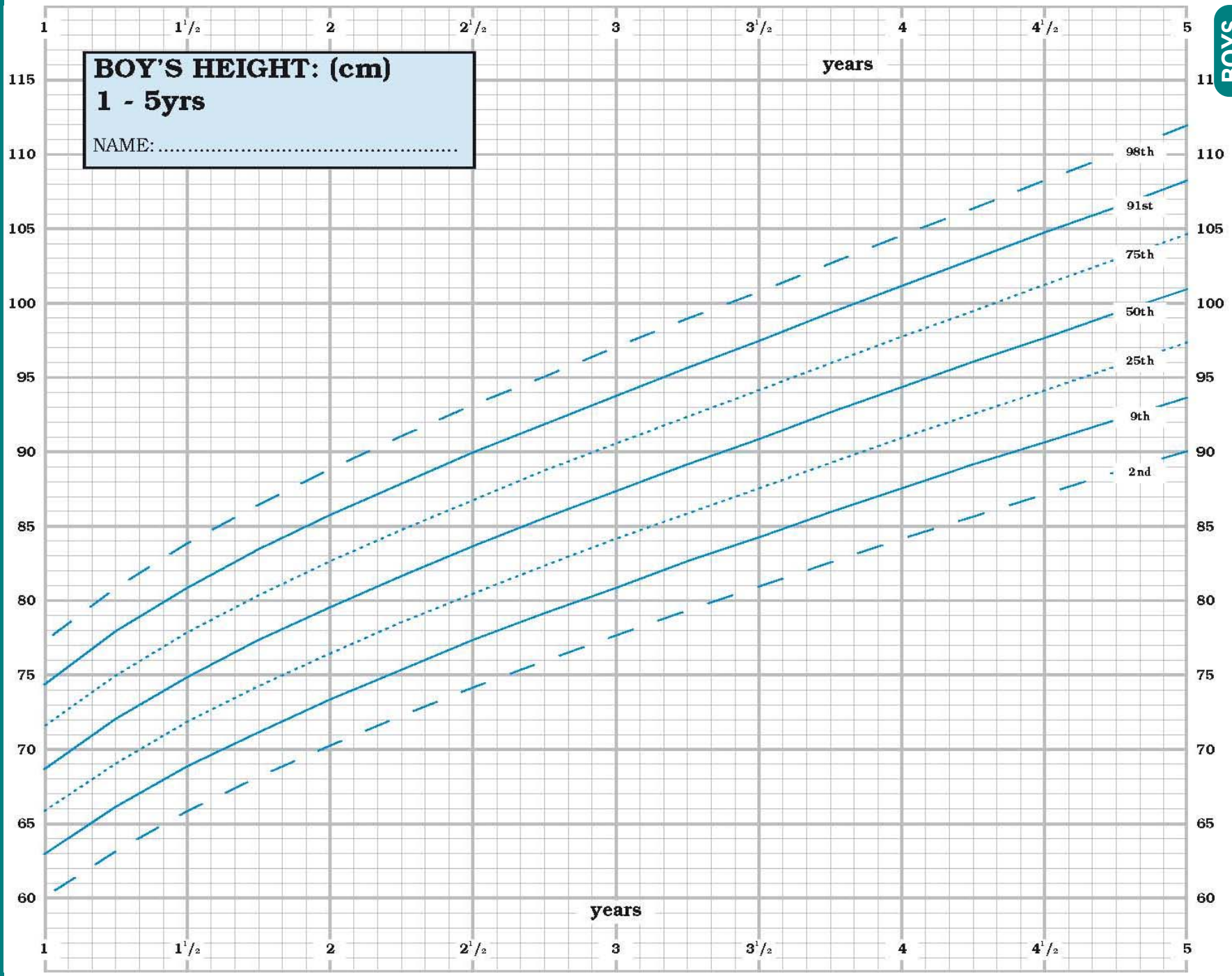
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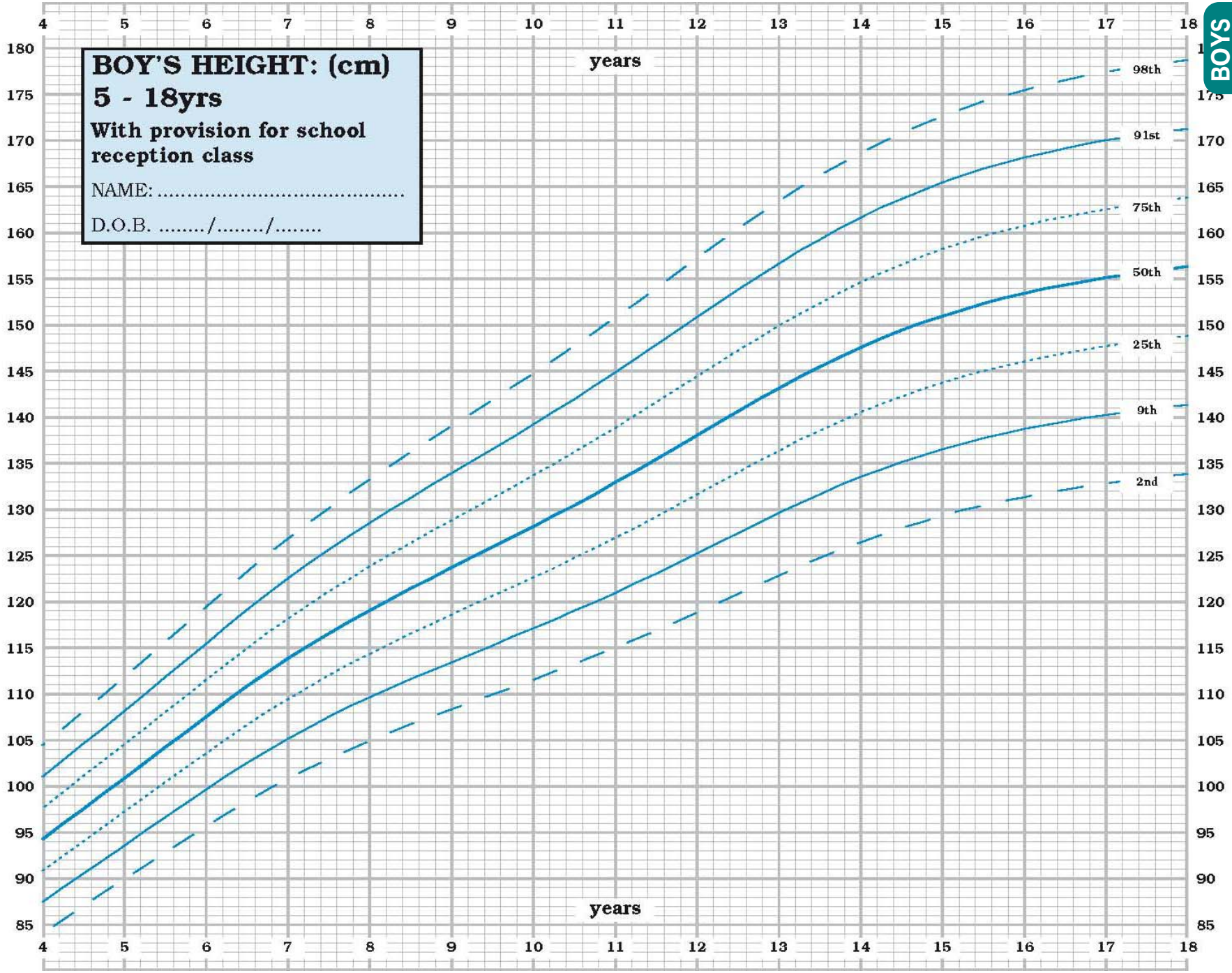
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5 - 18yrs
With provision for school reception class
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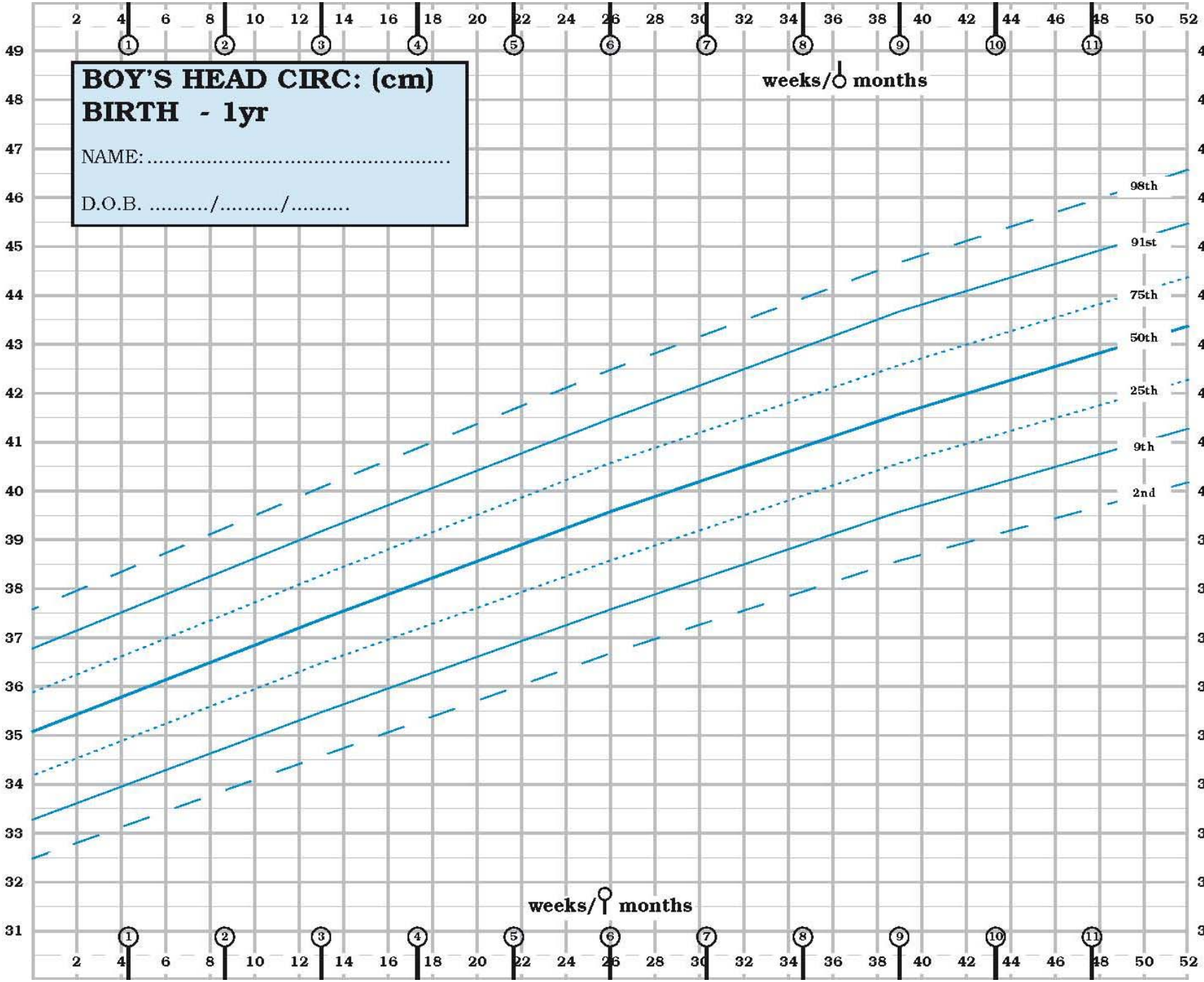
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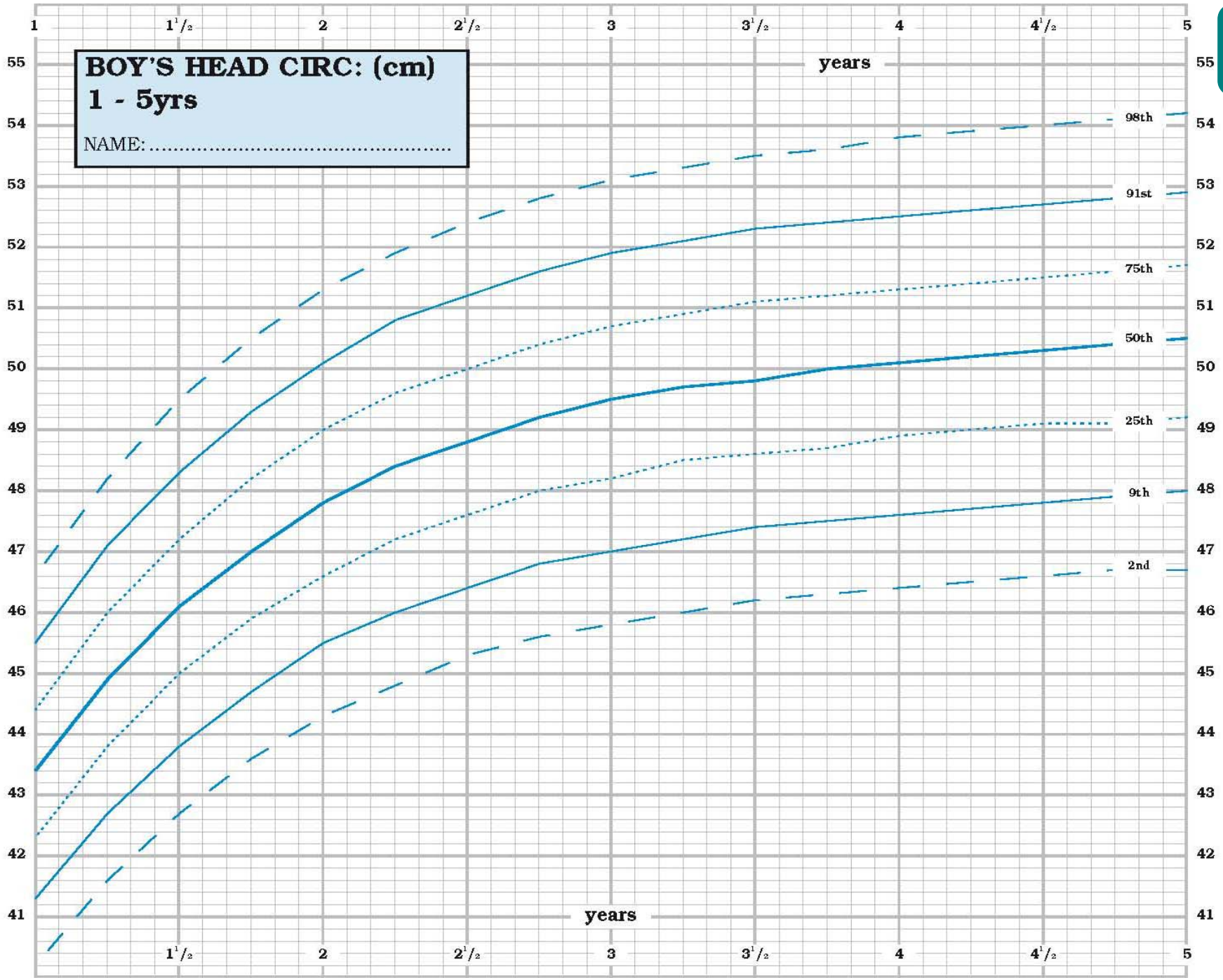
BOY'S HEAD CIRC: (cm)
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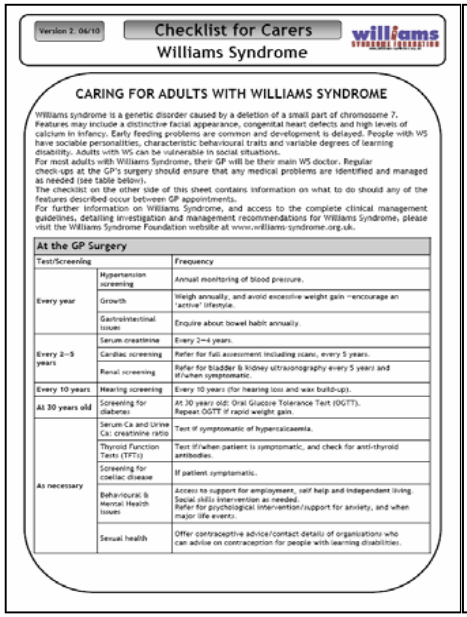
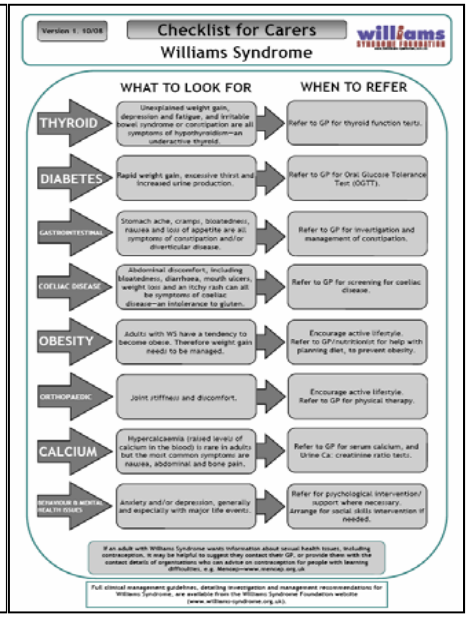
Other Resources


Checklist for Carers
Williams Syndrome

CARING FOR ADULTS WITH WILLIAMS SYNDROME

Williams syndrome is a genetic disorder caused by a deletion of a small part of chromosome 7. Features may include a distinctive facial appearance, congenital heart defects and high levels of calcium in infancy. Early feeding problems are common and development is delayed. People with WS have sociable personalities, characteristic behavioural traits and variable degrees of learning disability. Adults with WS can be vulnerable in social situations.
For most adults with Williams Syndrome, their GP will be their main WS doctor. Regular check-ups at the GP's surgery should ensure that any medical problems are identified and managed as needed (see table below).
The checklist on the other side of this sheet contains information on what to do should any of the features described occur between GP appointments.
For further information on Williams Syndrome, and access to the complete clinical management guidelines, detailing investigation and management recommendations for Williams Syndrome, please visit the Williams Syndrome Foundation website at www.williams-syndrome.org.uk.

At the GP Surgery		
Test/Screening		Frequency
Every year	Hypertension screening	Annual monitoring of blood pressure.
	Growth	Weigh annually, and avoid excessive weight gain —encourage an 'active' lifestyle.
	Gastrointestinal issues	Enquire about bowel habit annually.
Every 2–5 years	Serum creatinine	Every 2–4 years.
	Cardiac screening	Refer for full assessment including scans, every 5 years.
	Renal screening	Refer for bladder & kidney ultrasonography every 5 years and if/when symptomatic.
Every 10 years	Hearing screening	Every 10 years (for hearing loss and wax build-up).
At 30 years old	Screening for diabetes	At 30 years old: Oral Glucose Tolerance Test (OGTT). Repeat OGTT if rapid weight gain.
As necessary	Serum Ca and Urine Ca: creatinine ratio	Test if symptomatic of hypercalcaemia.
	Thyroid Function Tests (TFTs)	Test if/when patient is symptomatic, and check for anti-thyroid antibodies.
	Screening for coeliac disease	If patient symptomatic.
	Behavioural & Mental Health issues	Access to support for employment, self help and independent living. Social skills intervention as needed. Refer for psychological intervention/support for anxiety, and when major life events.
	Sexual health	Offer contraceptive advice/contact details of organisations who can advise on contraception for people with learning disabilities.


Checklist for Carers
Williams Syndrome

WHAT TO LOOK FOR / WHEN TO REFER

THYROID — Unexplained weight gain, depression and fatigue, and irritable bowel syndrome or constipation are all symptoms of hypothyroidism—an underactive thyroid. → Refer to GP for thyroid function tests.

DIABETES — Rapid weight gain, excessive thirst and increased urine production. → Refer to GP for Oral Glucose Tolerance Test (OGTT).

GASTROINTESTINAL — Stomach ache, cramps, bloatedness, nausea and loss of appetite are all symptoms of constipation and/or diverticular disease. → Refer to GP for investigation and management of constipation.

COELIAC DISEASE — Abdominal discomfort, including bloatedness, diarrhoea, mouth ulcers, weight loss and an itchy rash can all be symptoms of coeliac disease—an intolerance to gluten. → Refer to GP for screening for coeliac disease.

OBESITY — Adults with WS have a tendency to become obese. Therefore weight gain needs to be managed. → Encourage active lifestyle. Refer to GP/nutritionist for help with planning diet, to prevent obesity.

ORTHOPAEDIC — Joint stiffness and discomfort. → Encourage active lifestyle. Refer to GP for physical therapy.

CALCIUM — Hypercalcaemia (raised levels of calcium in the blood) is rare in adults but the most common symptoms are nausea, abdominal and bone pain. → Refer to GP for serum calcium, and Urine Ca: creatinine ratio tests.

BEHAVIOUR & MENTAL HEALTH ISSUES — Anxiety and/or depression, generally and especially with major life events. → Refer for psychological intervention/support where necessary. Arrange for social skills intervention if needed.

If an adult with Williams Syndrome wants information about sexual health issues, including contraception, it may be helpful to suggest they contact their GP, or provide them with the contact details of organisations who can advise on contraception for people with learning difficulties, e.g. Mencap—www.mencap.org.uk

Full clinical management guidelines, detailing investigation and management recommendations for Williams Syndrome, are available from the Williams Syndrome Foundation website (www.williams-syndrome.org.uk).

- **Williams Syndrome Review Checklist**

This is aimed at carers involved in the care of adults with Williams Syndrome.
It clearly states which tests and what screening should be undertaken at the GP surgery, and how often.
It also lists symptoms which may present between GP appointments, and what to do should they occur.
The checklist is currently being finalised, and will be available from Kay Metcalfe, Consultant Geneticist at St Mary's Hospital in Manchester (kay.metcalfe@cmft.nhs.uk) when it is completed.

- **Orphanet (www.orpha.net)**

Orphanet is an online database of rare diseases and related services provided throughout Europe. It contains information on over 5,000 conditions, including Williams Syndrome, and lists specialised clinics, diagnostic tests, patient organisations, research projects, clinical trials and patient registries relating specifically to Williams Syndrome.

Resources for Patients

- **Personal Health Record for Williams Syndrome (Blue Book)**

All babies in the UK are issued with a red book to record their health, growth and development. We have previously designed similar Blue Books for several rare conditions requiring multi-disciplinary management, including 22q11 Deletion Syndrome, Achondroplasia and Neurofibromatosis Type 1.
As part of this project a Blue Book has been designed for people with Williams Syndrome.
The primary aim of the Blue Book is to empower patients and their families, giving them more information about and ultimately more control over their health. It will also benefit the healthcare professionals involved in managing these patients, by facilitating inter-speciality communication, educating non-specialists and allied healthcare professionals, providing a readily accessible summary 'snapshot' of a patient's condition, and they can also be used as a tool for clinical audit and research.
They are available from the Williams Syndrome Foundation (see below)

- **The Williams Syndrome Foundation UK (www.williams-syndrome.org.uk)**

williams SYNDROME FOUNDATION
www.williams-syndrome.org.uk

The Williams Syndrome Foundation is run for parents by parents. They aim to be the first point of contact for individuals with Williams Syndrome, their families, and professionals needing support and information regarding the Syndrome.
The Foundation actively supports research into the educational, behavioural, social, scientific and medical aspects of the Syndrome, and seeks to organise their financial and personnel resources so as to achieve their mission on a sustainable basis.

Acknowledgements

- **The Williams Syndrome Guideline Development Group**
  Dr Jane Ashworth, Dr Susmito Biswas, Professor Bruno Dallapiccola, Dr Mark Dalzell, Dr Jane Deal, Professor Dian Donnai, Pam Griffiths, Dr Kay Hood, Professor Pat Howlin, Dr Ed Ladusans, Dr Ralph MacKinnon, Dr Josephine Marriage, Dr Neil Martin, Dr Kay Metcalfe, Dr Zulf Mughal, Dr Ramanlal Patel, Dr Alison Pike, Dr Christopher Stinton, Kate Strong, Dr Rajat Verma, Dr Mike Wolfman

- **DYSCERNE: A Network of Centres of Expertise in Dysmorphology (www.dyscerne.org)**

- **The Williams Syndrome Foundation (www.williams-syndrome.org.uk)**
  Regional Coordinators
  Professional Advisory Panel

- **Nowgen—A Centre for Genetics in Healthcare (www.nowgen.org.uk)**

These guidelines were produced thanks to funding from DYSCERNE: A Network of Centre of Expertise for Dysmorphology (funded by the European Commission Public Health Executive Agency (DG Sanco) Project: 2006122), and the Williams Syndrome Foundation UK.

Document Title: Management of Williams Syndrome: A Clinical Guideline
Version: 1
Created: 08/05/2009
Reviewed: 02/10/2009
Review Date: 01/06/2010

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