National Screening Committee

Child Health Sub-Group Report
Growth Disorders

September 2004

Growth Disorders

The condition

Growth disorders. There are many conditions that cause abnormally slow or fast growth in weight, height/length and head circumference. Slow weight gain in infancy ("faltering growth") is common. It may indicate serious diseases or parental management problems, but is often just a normal variation. Many medical disorders affect growth in height but there are very few conditions that present *only* with poor or very fast growth – the two important ones are Turner's syndrome (TS) and growth hormone deficiency (GHD). Abnormal growth in head circumference is rarely an isolated phenomenon.

Screening and benefits of screening

Screening involves taking measurements of head circumference, height or weight and plotting the result on a chart. This latter is essential for interpretation. The benefits of the process were considered in detail at a meeting in Coventry in 1998. Weight monitoring in infancy was thought not to fulfil the criteria for screening. Proposals for good clinical practice are set out in a paper by Wright.

It was specified that children should be weighed when they attend for immunisation or routine health checks at ages agreed as part of the child health programme.
With regard to height, a distinction was made between screening by a single height measurement and screening by monitoring growth over time to detect abnormal rates of growth – the monitoring of height velocity. A single height measurement, with cut-off at the 0.4 centile, at or around the age of starting school, was accepted as a screening test.

Previous recommendations had suggested that the crossing of centile lines on the growth chart could be used as a means of detecting abnormal growth if done over a period of time. However, the imprecision of measurement, and the variability in

growth rates, together supported the view that height velocity monitoring is not a practical means of screening.

Weight should also be checked at school entry. Although it is not a screening test, height and weight together are valuable items as part of a core public health data set.

Number of adverse events prevented each year in the UK by screening

Effective screening for short stature using the 0.4 centile should identify at least half of all previously missed cases of GHD and of TS, the two main target disorders. A delayed diagnosis can be regarded as an adverse event as it may result in reduced final height and psychological trauma, especially in TS.

These would benefit from earlier intervention leading to increased adult height and, in the case of TS, there are probably psychological advantages as well. There are no precise data but perhaps 40 cases of TS and double that number of GHD might be identified.

Sensitivity and specificity

Weighing and measuring have poor sensitivity and specificity in terms of identifying disease and normality. Measuring, plotting and interpreting the results are not simple. Many babies who have faltering growth are normal. Much harm can be done if staff do not understand the principles of how growth charts work, normal variation and regression to the mean. The problems with height monitoring are to do with quality, accuracy, unavoidable measurement error and low yield of genuinely new cases.

Economic aspects

Weighing and plotting are part of normal practice and even if they were discontinued, parents would still bring their babies to be checked. A single height screening measurement would have a measurable cost, which would depend on whether it was integrated into other routine health care activities. If the measurement were to be done by a practice nurse, for example when the child attends for the pre-school booster, or by a technician or school nurse at the same time as a hearing or vision

test, the cost would be negligible. If done separately as an isolated item it would be necessary to carry out a small observational study to assess the time needed. The value of height and weight measures would be greatly enhanced if the data were to be added to a core data set for child public health. However, the cost is *not* the reason for rejecting height velocity monitoring as a screening test – the reason is the poor test performance.

Disorders of growth – short stature due to growth hormone deficiency (GHD) and Turner's syndrome (TS) in particular

The condition

1. **The condition should be an important health problem.** Untreated these conditions can give rise to short stature in adulthood and some associated problems which may be treated. The deficit in stature may be reduced by early treatment. The disorders are moderately common -1:3000 for GHD; 1:2500 girls for Turner's syndrome.

2. **The epidemiology and natural history of the condition, including development from latent to declared disease, should be adequately understood and there should be a detectable risk factor, or disease marker and a latent period or early symptomatic stage.**
YES

3. **All the cost-effective primary prevention interventions should have been implemented as far as practicable.**
N/A

4. If the carriers of a mutation are identified as a result of screening the natural history of people with this status should be understood, including the psychological implications.
N/A

The test

5.There should be a simple, safe, precise and validated screening test.
Height measuring is safe and simple in principle. However, in practice, it is often not accurately reproducible.

6. The distribution of test values in the target population should be known and a suitable cut-off level defined and agreed.

Measurement imprecision and error are well known. Height charts are available.

7. The test should be acceptable to the population.
YES

8. There should be an agreed policy on the further diagnostic investigation of individuals with a positive test result and on the choices available to those individuals.
A nationally agreed protocol has yet to be agreed.

9. If the test is for mutations the criteria used to select the subset of mutations to be covered by screening, if all possible mutations are not being tested, should be clearly set out.

N/A

The treatment

10. There should be an effective treatment or intervention for patients identified through early detection, with evidence of early treatment leading to better outcomes than late treatment.
YES for some conditions.

11.There should be agreed evidence based policies covering which individuals should be offered treatment and the appropriate treatment to be offered.
YES for some conditions

12.Clinical management of the condition and patient outcomes should be optimised by all health care providers prior to participation in a screening programme.
NO

The screening programme

13.There must be evidence from high quality Randomised Controlled Trials that the screening programme is effective in reducing mortality or morbidity.

Utah study shows new cases identified and treated with benefits

Where screening is aimed solely at providing information to allow the person being screened to make an "informed choice" (e.g. Down's syndrome, cystic fibrosis carrier screening), there must be evidence from high quality trials that the test accurately measures risk. The information that is provided about the test and its outcome must be of value and readily understood by the individual being screened.

N/A

14.There should be evidence that the complete screening programme (test, diagnostic procedures, treatment/intervention) is clinically, socially and ethically acceptable to health professionals and the public.

YES

15.The benefit from the screening programme should outweigh the physical and psychological harm (caused by the test, diagnostic procedures and treatment).

YES, but only if protocols observed.

16.The opportunity cost of the screening programme (including testing, diagnosis, treatment, administration, training and quality assurance) should be economically balanced in relation to expenditure on medical care as a whole (i.e. value for money).

This exercise has not been conducted.

17.There should be a plan for managing and monitoring the screening programme and an agreed set of quality assurance standards.

These could be drawn up.

18.Adequate staffing and facilities for testing, diagnosis, treatment and programme management should be made available prior to the commencement of the screening programme.

Probably possible, depending on the referral criteria..

19.All other options for managing the condition should have been considered (e.g. improving treatment, providing other services), to ensure that no more cost effective intervention could be introduced or current interventions increased within the resources available.

N/A

20.Evidence based information, explaining the consequences of testing, investigation and treatment, should be made available to potential participants to assist them in making an informed choice.

These could be drawn up.

20. Public pressure for widening the eligibility criteria for reducing the screening interval, and for increasing the sensitivity of the testing process, should be anticipated. Decisions about these parameters should be scientifically justifiable to the public.

N/A.

21. Public pressure for widening the eligibility criteria for reducing the screening interval, and for increasing the sensitivity of the testing process, should be anticipated. Decisions about these parameters should be scientifically justifiable to the public.

As they are continuous variables and there is no cut-off that reliably distinguishes 'cases' that will benefit from treatment from those who won't, there is likely to be continuing debate. Whatever cu-off were to be chosen would need to be justified on astatistical basis.

22.If screening is for a mutation the programme should be acceptable to people identified as carriers and to other family members.

N/A

Summary – recommendations

- Weight monitoring in infancy is not screening but it is potentially useful provided the weight is correctly interpreted.
- Babies should be weighed on request and when they have contacts with the health service for other reasons.
- Staff must be trained in how to interpret growth charts and must understand the mathematical difficulties in determining whether or not a baby is truly failing to thrive.
- Height: it is good practice to check and plot height opportunistically and in any chronic health problem. The 0.4 centile line in the chart is the cut-off for referral or action. However, the evidence does not justify defining *repeated* measurements as a screening programme, either as a series of individual measurements or as a means of deriving a velocity.
- Children should have their height and weight measured at around the time of school entry and the 0.4 cut-off for height should be used to initiate referral. The evidence suggests that this would have a low but useful yield and should be treated as a screening test.
- Parental height should not be taken into account when assessing these screening measurements at primary care level.
- The evidence is firmly against the use of serial height measurements with the aim of identifying abnormal growth velocity, as a routine screening procedure.
- Height and weight at school entry should be part of the Essential Core Data Set for child public health.
- Quality monitoring. This requires training of clinical staff both for screening and for receiving referrals; data collection using IT systems and analyses of measurements to monitor error, imprecision and interventions; central monitoring of cases found.

Sources of information

Literature reviews; meeting of experts at Coventry 1998 – the Coventry consensus. (Hall DM. Growth monitoring. Arch Dis Child. 2000 Jan;82(1):10-5.) Conclusion supported by all but one member of the meeting.

Wright CM. Identification and management of failure to thrive: a community perspective. Arch Dis Child. 2000;82(1):5-9.

Status of the recommendations

Debated and agreed in CHSG. Expert opinion. No formal RCTs, but published studies and simple modelling support the conclusions. N.B.: head circumference measurement, as a screening test has not been discussed in detail.

Consumer view

Conclusions accepted by parent organisation (Child Growth Foundation – sponsors of Coventry consensus meeting) though they would prefer extended programme of growth monitoring if evidence supported it. Both the Foundation and expert advice strongly support improved training for all staff and a video is in preparation.

Quality of evidence

II-1, II-2, II-3, III.

Strength of recommendations

Good evidence to support single height measure. Good evidence to reject height velocity monitoring.

Research agenda

1. Neither clinical evidence nor modelling support height velocity monitoring. However, the importance of early diagnosis is agreed. A comparative trial of monitoring versus current practice, using age of diagnosis of GHD or TS as outcomes, would not be feasible.

2. Age of diagnosis of GHD and TS might be a useful clinical outcome indicator. If established, it would offer a mechanism for monitoring progress in implementing good practice guidelines and effectiveness of training and referral practices.

3. The reasons for delay in diagnosis need further study. How much does this occur at the level of parent, primary care, or secondary care?