ENDOCRINE POTPOURRI – TOO SHORT, TOO FAT AND TOO SWEET...

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A potpourri of common paediatric endocrine problems which you may encounter in your clinical practice is discussed. With each problem, diagnostic pegs will be highlighted, with specifics of what you can do as the primary health care physicians, and what we might be able to offer at the tertiary level.

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• The obese child
• The child with diabetes mellitus
'TOO SHORT'

Growth is an essential and integral component of Paediatrics. It is a sensitive index of a child's health. Short stature may be the first indication of a disease state. More significantly, it may be a psychological disability, because short children may be teased mercilessly, bullied, or excluded from certain activities.

A common problem which you may encounter is "How short is too short?" A short child is arbitrarily defined as any child whose height is less than the 3rd percentile for his or her community.

WHAT YOU CAN DO:

As the primary health care physician, you can assess short stature by following these steps:

1. Exclude 'perceived short stature';
   a) Measure the child's height accurately
      Heights must be accurately measured. Inaccurately measured heights may cause unnecessary anxiety or may give a false sense of security to the parents. The gold standard for measuring heights is with the wall mounted stadiometer. However, these are very expensive, and usually only available at the tertiary hospitals. The wall mounted charts and floppy arm devices for measuring heights are a useful substitutes, but the correct techniques using these apparatus need to be appreciated.
      
      The child should be barefoot with the heels, buttocks and shoulders in contact with the wall. The heels should be placed together. The head should be in the Frankfort horizontal plane, a plane represented by a line between the lowest point on the margin of the orbit and the highest point on the margin of the auditory meatus. If the floppy arm device is used, it is very important that the scale arm must be level, and not dipping down or up.
      b) Plot the height on the relevant growth chart
      Heights need to be plotted on Singapore growth charts for boys and girls, and a short child is one who is less than the 3rd percentile for his or her age.
2. **Assess for familial short stature by determining the adjusted mid-parental height (target height):**

Parental heights need to be considered. There is a genetic component to short stature, and short parents beget short children. Familial short stature constitutes 40 percent of all cases of short children.

The genetic potential of a child can be assessed by considering the adjusted mid-parental heights. The difference between a male and female of 13 cm must be taken into account in the equation for the target height, or adjusted mid-parental height:

For a boy, the target height \( = \frac{F + (M+13)}{2} \)

For a girl, the target height \( = \frac{(F-13) + M}{2} \)

The target range will be the target height ± 7.5 cm for the 10th to 90th percentiles of the genetic potential. By plotting the target height and target range on the growth chart, a good estimate of the child's genetic potential can be made.

3. **Assess for constitutional growth delay:**

Another 40 percent of short children are late developers, and have constitutional growth delay. It is therefore also important to enquire if the parents had a history of delayed puberty. When did mother's menarche occur, and when did father's voice change?

4. **Assess for pathological short stature:**

Pathological short stature is a possibility after excluding perceived short stature and normal variant short stature. The greater a child deviates from the 3rd percentile, the higher likelihood that the child has a pathology.

Pathological short stature can be proportionate or disproportionate which may be due to a variety of short limb or short trunk dwarfs. Once a diagnosis of pathological short stature is made, the child should be referred to the tertiary hospital for an assessment. However, some spot diagnoses you can make include:
a) Turner syndrome:
The classical Turner syndrome has proportionate short stature, a stocky build, neck webbing, a broad shield chest, inverted or hypoplastic nipples, a wide carrying angle, short 4th and 5th metacarpals, and a trident hairline. All Turner girls must be examined carefully for a cardiac problem - especially to feel the pulses to exclude a coarctation of the aorta. One important point is that many Turner girls especially mosaic Turner syndrome, with a significant population of 46 XX cells, may look completely normal, apart from short stature. It is therefore mandatory for all short girls to have a chromosome karyotype performed.

b) Growth hormone deficiency:
The child with congenital growth hormone deficiency has severe short stature, is relatively plump, with a cherubic facies, and a high pitched voice.

c) Cushing's syndrome:
Excess steroids can cause short stature because they inhibit growth hormone production centrally, and affect the response of the growth plate. The phenotype is unmistakable in overt Cushing's syndrome with the Cushingoid facies, hirsutism, buffalo hump, violaceous striae, acne, and proximal myopathy. However, the pituitary adenomas may not be so evident, and the first sign may simply be excessive weight gain with poor growth.

5. Monitoring a child's growth and concept of height velocity:

If the child has perceived short stature, or normal variant short stature (familial short stature or constitutional growth delay), you can reassure the parents, and follow up the child by monitoring sequential heights. The child's growth is normal if he continues to grow parallel to the height curves. However, if he deviates and plateaus, a re-assessment is required.

In an equivocal situation, you may choose to continue monitoring the child's growth for another 3 to 6 months. While height is a static measurement, height velocity (measured as cm per year) is a critical parameter of the dynamic function of growth. If a child is growing more than the magical figure of 4 cm per year, or 2 cm in 6 months, his growth is acceptable, and he is unlikely to have any serious pathology.

However, some parents may insist on answers instead of waiting, and you may choose to refer the child to us at any point for a second opinion.
WHAT WE CAN DO:

A. **Diagnostically**, we can perform the following investigations:

1. X-ray left hand for bone age - A normal bone age usually excludes a pathology.

2. Chromosomal karyotype - This is the definitive test for Turner syndrome. The old system of doing a buccal smear for Barr bodies is not accurate, and has largely been superseded.

3. Growth hormone(GH) stimulation tests
   A variety of tests are available, as there is no single test which can reliably diagnose GH deficiency. The diagnosis of GH deficiency traditionally depends on at least 2 dynamic tests of GH stimulation. At NUH, we perform:
   a) The exercise GH stimulation test
   b) The arginine GH stimulation test
   c) The insulin hypoglycaemia GH stimulation test
   d) The glucagon GH stimulation test
   We can also check the free IGF-1 levels, and IGF binding proteins.

4. Other screening tests for systemic disease:
   a) Renal function tests, as renal tubular acidosis or chronic renal failure may have short stature as the initial presenting problem.
   b) Thyroid function tests - at NUH, we perform the free thyroxine levels, which supersede the total thyroxine level, and we have the third generation TSH assays, with marked improvement in accuracy.
   c) Radiological studies to exclude inflammatory bowel disease.
   d) MRI scanning of the hypothalamic pituitary region to exclude a tumour which causes panhypopituitarism.

B. **Specific therapy:**

   a) Once the pathology has been identified, we can initiate specific treatment to optimise the heights of these short children. The well established indications for GH therapy include GH deficiency and Turner syndrome.

   While GH is expensive, we can tap into the NUH Children's Assistance Plan Fund. Although this funding is limited, children who are desperately short and in need of GH can be partially subsidised for treatment. We also follow-up these children by measuring their heights every 3 to 6 monthly, adjusting their dose of GH commensurate with their increasing body surface area, and monitoring for potential complications of GH treatment.
b) Genetic counselling can be provided if the pathology diagnosed is inherited. The bone clinic has recently been commenced. This is a 3 monthly multi-disciplinary clinic for short children with bone dysplasias and rickets. It is attended by the paediatric endocrinologist, the geneticist, the paediatric nephrologist, the paediatric orthopaedic surgeon, the neurosurgeon (where appropriate), the physiotherapist and the medical social worker.

c) Social support services are available for Turner syndrome. The NUH Turner support group will be commencing in March 1998, and apart from the psychosocial support provided, literature and information on the latest research in Turner syndrome can be made available for children and adolescents with Turner syndrome.

FLOW CHART FOR SHORT STATURE

Is my child short?

Exclude perceived short stature
(a) Measure height accurately
(b) Plot on growth chart

$3rd$ percentile
‘Perceived’ short

< $3rd$ percentile
short

Exclude Familial Short Stature (FSS)
Target height/range?

Reassure
Monitor Ht
Velocity (HV)
3-6 monthly

FSS

CGD

HV <4 cm/yr

?Pathological Short Stature

REFER
"TOO FAT"

Childhood obesity is an increasing problem, and it is the second most common health problem in Singapore school children. With increasing affluence, there is an increased intake of dietary fat, associated with decreased physical activity.

Obesity is associated with increased morbidity and mortality. The complications include hypertension, hyperlipidemia, increased risk of coronary artery disease, type 2 diabetes, obstructive sleep apnea syndrome, degenerative joint disease, gallstones and the Pickwickian syndrome of obesity-hypoventilation syndrome. Obesity in childhood leads to approximately 30 percent of obesity in adulthood. Many fat adults were fat infants, and the natural history suggests that the obese child who becomes an obese adult will have more severe adult obesity than adults whose obesity began in adulthood. It therefore important to assess and intervene in childhood.

WHAT YOU CAN DO:

As the primary health care physician, you can:

A. Assess the degree and severity of obesity in several ways:

1. Weight for age - the same principle of charting weight percentiles, as with height measurements.

2. Weight for height charts - A child is considered obese if he is >20 percent above his ideal weight for height, and he is morbidly obese if he is >100 percent above his ideal weight for height.

3. Body mass index (BMI) - can be calculated based on the formula:

\[
\text{BMI} = \frac{\text{Weight (kg)}}{\text{[Height (m)]}^2}
\]

Garrow's classification correlates the BMI with health risk, such that:

- BMI 25-30 kg/m²: low risk
- BMI 30-35 kg/m²: moderate risk
- BMI 35-40 kg/m²: high risk

B. Identify pathological fatness and refer for a full assessment:
a) 'Fat child' syndromes - Prader Willi syndrome, Lawrence Moon Biedl syndrome.

b) Cushing's syndrome - may commence initially with excessive weight gain, usually with concomitant poor height gain. The younger children tend to have adrenal tumours, while the older children usually have pituitary adenomas.

c) Severe morbidity obesity may be due to a hypothalamic tumour which needs to be excluded.

C. If the child has exogenous obesity due to dietary indiscretion and lack of exercise, you can:
   a) Highlight the problem to the parents and the child.
   b) Take a good dietary history and provide simple dietary advice with recommendations on exercise.
   c) Monitor the child's weight for 1 to 3 months.
   d) Reinforce that permanent changes can only be achieved if there is a significant change in family lifestyles. Often the entire family must partake of a healthier lifestyle.
   e) Recalcitrant cases who continue to gain weight despite advice can be referred to us for assessment and management.

WHAT WE CAN DO:

1. Objective assessment of obesity:
   We can perform a body fat measurement using:
   a) The fat calipers
   b) The dual energy X-ray absorptiometer (DEXA) which gives a detailed objective assessment of the percentage body fat, lean body mass, and bone mineralisation, as a good baseline for subsequent comparison.

2. Exclude a pathological cause:
   a) Cushing's syndrome can be excluded by measuring the diurnal cortisol production at 8 am and 12 midnight, performing a 24 hour urine for free cortisol assessment, and a screening overnight dexamethasone suppression test. If hypercortisolism is clearly present, then a low dose and high dose dexamethasone suppression test will be performed.
   b) Exclude other endocrine problems such as hypothyroidism and growth hormone deficiency.
   c) Exclude a hypothalamic lesion by MRI scan of the brain.

3. Dietary intervention:
The paediatric dietician will assess the intake of the child and family, and design an individualised diet plan with the main principles of reduction in fat intake. A good understanding of food groups and nutrition helps in the choice of food alternatives. The techniques of cooking are also emphasised, especially the avoidance of fried foods, and resorting to steamed and boiled foods.

4. Exercise programme:

The exercise physiotherapist can design a programme for the children to ensure daily regular exercise in the form of some aerobics. Very young children do not generally need a formal exercise programme as active play, preferably outdoors should create sufficient activity for energy balance. Sedentary activities such as watching TV can be replaced by more physical activity.

5. Ward admission for weight loss

In recalcitrant cases, we admit the obese children, so that an enforced diet and exercise programme with education can be provided in a controlled environment. We plan to admit the obese children together during the school holidays, so that they can be counselled together, and hopefully motivate one another through healthy competition. However, for any weight loss programme to succeed, permanent changes in lifestyle are essential. Hence the need for continued follow-up, encouragement and on-going advice with regards to behaviour modification.

6. Behaviour modification:

Parental involvement is critical for any successful weight reduction programme, and we usually counsel the families to modify their lifestyles in terms of dietary practices and activities.

7. Screening for complications of obesity:

In the obese child, we also perform baseline tests which include the fasting serum cholesterol, HDL-cholesterol, LDL cholesterol, and serum triglycerides. We screen for possible type 2 diabetes with glycosylated hemoglobin levels, and a formal oral glucose tolerance test, as well as the thyroid status of the children. Any complications can be monitored with weight reduction, and treated if necessary.
FLOW CHART FOR OBESITY

Is my child obese?

Initial assessment
(a) Weight for age
(b) Weight for height
(c) BMI = \( \frac{\text{Wt (kg)}}{[\text{Ht (m)}]^2} \)

Identify pathological obesity
(a) ‘Fat child’ syndrome
(b) Cushing’s syndrome
(c) Morbid obesity (>100% above ideal wt for ht)
? Hypothalamic tumour

Yes
No

? Exogenous obesity

REFER

Manage
(a) Diet, exercise, lifestyle recommendations
(b) Monitor weight for 1-3 months

Weight gain
Static weight
(with hypertension or other complications)

Weight loss
Give due encouragement and support

'TOO SWEET'

Childhood diabetes is increasing in Singapore from 0.04 per 100 000 school age children less than 12 years in 1981 to 3.8 per 100 000 school age children in 1994. At the NUH Paediatrics Department, we have 47 families with diabetes, of which 94 percent are type 1 and 6 percent type 2. Our philosophy in management is to provide holistic care and psychological support through a multidisciplinary paediatric diabetes team which includes the paediatric diabetologists, the paediatric diabetes nurse educators, the dieticians,
podiatrist, medical social worker, as well as the closely linked community support services.

WHAT WE DO:

1. All newly diagnosed diabetics and their families are given a full diabetes education and counselling service while in the wards by the paediatric diabetologist, diabetes nurse educators and dietician. Much time and effort is invested at initial diagnosis because we feel that the cornerstone of good diabetes management lies in a solid foundation created through diabetes education. Since diabetes is chronic, and it requires daily insulin injections, the parents and children eventually must function as their own doctors. The more they understand about diabetes, the better the control.

   The education sessions cover:
   a) The pathogenesis of diabetes
   b) Techniques of injections and blood glucose monitoring
   c) Hypoglycemias and their management
   d) Sick days and hyperglycemias
   e) Complications

2. Coping with diabetes is an on-going task. Follow-up clinics are held at the NUH Diabetes Centre where the glycosylated hemoglobin levels are assessed, with results known within 10 minutes. Apart from seeing the doctor, the families are also attended to by the diabetes nurse educators, the dietician, and the community social support service. The post-clinic conference allows input from all participants of the diabetes team, a discussion of how families are coping, and decisions made with regards to the strategy of management.

3. Annual diabetes camps have also been organised since 1995. These camps provide an opportunity for the diabetic children to interact with one another. It encourages the children to develop the confidence to live as normal children, and is a good opportunity for the staff to get to know the families better.

WHAT YOU CAN DO:

1. With an increasing incidence of diabetes, it is important to be astute in diagnosing a child with diabetes. The initial presentation can masquerade as hyperpnea in DKA, or prolonged ill-health, apart from the usual polydipsia and polyuria. "If in doubt, check it out" by performing a quick and simple urine test for sugar.

2. For known diabetics, the primary health care physician can reinforce the need for compliance to dietary regimens and monitoring. Psychosocial support can be
provided at this level, and any problems or complications detected early can be referred to us for further management.

3. The primary physician can continue to manage common ailments in diabetes. However, one needs to be acquainted with:
   a) The use of glucose free medications - tablets, or sorbitol based syrups.
   b) The principles of sick day management which include ensuring adequate hydration, frequent blood sugar monitoring, and checking for urine ketones. If unsure, these diabetic children can be referred to the tertiary care hospitals.

4. Acute emergencies such as:
   a) Hypoglycemias and their management, in particular, the use of intramuscular glucagon for a hypoglycaemic coma or seizure.
   b) Children in diabetic ketoacidosis must be referred to a tertiary hospital for management.

FLOW CHART FOR CHILDHOOD DIABETES

Does my child have diabetes?

• Urine test for sugar
• Check fasting/postprandial blood glucose level

No
Reassure

Yes
REFER for stabilisation and DM education, counselling

Follow-up with primary health care physician

Reinforce compliance to diet, monitoring

Psychosocial support

Manage emergencies:
(a) Hypoglycemia mild to severe
(b) DKA - admit to hospital

Manage common ailments:
(a) Glucose free medications
(b) Sick days:
   i. Adequate hydration
   ii. Monitor BGL
   iii. Check urine ketones