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JPCH Short Case: Dysmorphology (title page)

SHORT CASE: AN APPROACH TO THE CHILD WITH DYSMORPHIC FEATURES

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SHORT CASE ARTICLE

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CONFLICTS OF INTEREST
There are no conflicts of interest to declare.

RELATIONSHIPS
Dr Kaufman is the course convenor of Genetics For Trainees, an independent educational course for paediatric and physician trainees. Dr White is a lecturer at the Genetics For Trainees course.

Dr White is a lecturer for hospital based genetics teaching for paediatric basic trainees. Dr Kaufman has previously arranged hospital based exam tutorials for paediatric basic trainees in his former role as Deputy Chief Resident Medical Officer, 2014.

SHORT CASE: AN APPROACH TO THE CHILD WITH DYSMORPHIC FEATURES

Like all short cases, the aim of the dysmorphology examination is to reach a focused list of differential diagnoses, which requires a systematic and structured examination. Have a general approach in mind, but tailor what is appropriate for the patient in front of you. Many of these principles will also apply in the long case physical examination, and in clinical practice. Comment on findings either as you go or at the end, then summarise your main findings before presenting a concise and considered differential diagnosis.

INTRODUCTION TO THE DYSMORPHOLOGY SHORT CASE

Show the child, parent and examiner that you are a kind, sensitive and considerate doctor. In any description of a child's appearance, use appropriate, neutral language. Words or phrases that could be used include 'distinctive appearance', 'different in appearance to parents', 'feature' or 'trait'. Avoid jargon or judgemental terms like 'dysmorphic', 'deformed' or 'abnormal'.

Be systematic and divide the examination into sections: start with a general inspection, then move from hands to head to face to torso to legs. Know what you are looking for and how to describe it. If in doubt, fall back on the '3S approach': Size, Shape, and Symmetry. Be flexible: depending on how the child is feeling you may need adapt your plan and stand back, change track, and just comment on what you observe.

GENERAL OBSERVATIONS

Start with your general observations, taking time to observe the child's appearance. Many trainees use a pneumonic such as 'IHUGVIDEP' to ensure they do not miss any key findings.

INTRODUCE: introduce yourself to the patient/family
HANDS: wash your hands before examining if you have not already done so
UNWELL/WELL: does the child seem sick or well?

GROWTH: measure and plot the weight, height and head circumference, and note the child's body habitus and proportions
VITALS: ask at the start for vital signs only if it seems indicated, better to check them yourself later in context
IATROGENIC: look for a feeding tube, mobility aids, toys that may give a clue regarding level of cognition, or other devices
DYSMORPHISM: what are your first impressions of the child's appearance; does he/she resemble their parents?
EXPOSE: if the child is co-operative and when appropriate, remove their shirt to look for scars, lines and skin lesions
PAIN: is the patient comfortable to be examined, and where is best to examine them: on the bed, chair, or parent's lap?

HANDS & UPPER LIMBS

Next move to the hands. Describe the hand size, shape and symmetry. Look at the nails, finger length and shape, and palmar creases. Look at the arms, and the segment proportions, commenting on any asymmetry or joint hypermobility.

HEAD

Look at the child's head, and its size, shape, fontanelles, sutures and hair (quality, quantity). Focus on the face and its shape, symmetry, coarseness, and divide the face into thirds.

For the upper third, assess the position of the hairline and the shape of the forehead. The eyes are an important part of the examination. Start externally and work inwards noting the eyelids, palpebral fissures (length, slant), spacing, sclera, iris and pupils. Then move to the midface. Comment on the nose and its size, shape, nasal bridge, tip, nostrils and philtrum. The ears are also important and note their size, shape, position, rotation, any skin tags or creases. For the lower third of the face, look at the jaw, lips, philtrum, gums, teeth, palate, tongue, uvula, and for the presence of any midline defects. If you find one midline defect, remember to look for others.

TORSO & SKIN

Look specifically at the skin and any scars, neurocutaneous stigmata, or pigmentation. Look for the presence of webbing or skin folds of the neck.

Look from in front and behind at the back and chest, specifically the spine, sternum and nipples. Listen to the heart sounds as cardiac anomalies are a feature of many syndromes with dysmorphic features. Examine the abdomen for scars, hernia or the presence of organomegaly.

LOWER LIMBS & FEET

Now move down to look at the legs and their segment proportions, comment as for the arms on any asymmetry or hypermobility. Look at the feet carefully and the nails, toes, webbing, and the foot size and shape (flat, curved, asymmetrical). Comment on footwear and mobility aids, and gait if the child is mobile.

GENITALIA (consider if appropriate)

It is often not appropriate to examine the child's genitalia in an exam scenario. Consider commenting instead on what you might look for on examination, and tell the examiner that you will not do this in the exam today. For the genitalia observe the phallus, scrotum and testes (size, development, descent) in boys, the labia in girls, and pubertal development in both. Note the position of the anus.

OTHER

Consider a targeted examination of other specific areas looking for relevant positive and negative findings based on what you have found so far. This might include checking vision and hearing, or fundoscopy, in some cases.

Think about the child's interaction and co-operation during the examination, and does this give you any clues about their development, cognition and neurology. What has their conversation and play been like? Were any characteristic behaviours, such as hand-flapping, observed?

If the parents or siblings are present in the room, look for clues about potential inheritance patterns, or possible non-syndromic familial features.

SUMMING UP

Hopefully by now you can summarise your salient findings. Are there any conditions that could tie your findings together, or provide a differential diagnosis? What differentiating features add weight to your differentials? Mention also any relevant negative findings that assist you prioritising your list of differentials. For example in the child with short stature where you have also noted a webbed neck, consider Turner syndrome in a girl or Noonan syndrome in a boy or girl; for a child with short stature with a thumb/radial anomaly, consider Fanconi Anaemia; for a child with short stature and elfin facies consider Williams syndrome.

Complete your examination in around 10 minutes to allow time for presentation of your findings and discussion. Consider what genetic and other tests you might do to assist in finding a unifying diagnosis. If in doubt, go for microarray as the aneuploidies and microdeletion/duplication syndromes will be detected by this method.

Don't panic: finding and describing features accurately is more important than being certain of the exact diagnosis. The diagnosis may be obscure, or even unknown. Be open, but avoid making judgements especially in front of the child and parents. As always, be mindful to use sensitive language to describe your findings, even when dashing through the dysmorphology short case.

Table 1: Key features on examination and syndromes to consider

KEY FEATURE	SYNDROMES TO CONSIDER
Short stature	Turner, Noonan, Russell Silver, Williams, skeletal dysplasias
Tall stature	Marfan, Homocystinuria, Klinefelter, Sotos
Obesity	Prader Willi, Bardet Biedl
Pubertal delay	Turner, Klinefelter
Aortic stenosis	Williams
Pulmonary stenosis	Noonan, Williams, Alagille
Conotruncal/AVSD	22q11 deletion, trisomy 21
Aortic dilatation	Marfan, Ehlers Danlos
Radial ray anomalies	Fanconi Anaemia, VACTERL, TAR, Blackfan Diamond
Deafness	Goldenhar, CHARGE, Waardenburg, Treacher Collins, Alport, NF-2
Cleft lip/palate	22q11 deletion (cleft palate), Stickler
Café au lait macules	NF-1, Fanconi Anaemia, McCune-Albright