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Approach to Dysmorphic Features

Developed by Evelyn Armour and Dr. Melissa MacPherson for PedsCases.com.
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Introduction:

Hello everyone, thank you for tuning into this episode of PedCases podcasts. My name is Evelyn Armour, and I am a medical student at the University of Alberta. This podcast was created under the guidance of Dr. Melissa MacPherson, a medical geneticist at the University of Alberta in Edmonton. The aim of this episode is to present the listener with an approach to recognizing and investigating dysmorphic features in the infant or child, whether that is in family practise, a pediatric clinic or in the delivery suite.

By the end of this episode, listeners should be able to:

1. Identify dysmorphic features and explain their significance
2. Discuss salient aspects in the history and physical exam,
3. Generate a basic differential diagnosis for a child with dysmorphic features,
4. Outline key investigations
5. And lastly, Decern the criteria for an appropriate referral to genetics.

Case Study

Before we dive in, I'd like to present a case that can help guide our framework for an approach to dysmorphic features. Roger is a 6-month-old male who was referred to you, a community pediatrician, for low muscle tone, failure to thrive, and a heart murmur at his recent well-child check-up. According to the referral note, Roger's parents state that he has always been a "small baby" and struggles to gain weight, despite regular feeds. After breastfeeding initially, Rogers' parents noticed he was having issues latching to the breast and have tried switching to formula, but he is tiring easily on the bottle and is taking a long time to feed. He appears to have good social skills with good eye contact and frequent smiles. Roger's parents describe him as "floppy" when held. They note he is not rolling over, and he cannot sit with support. Stay tuned to hear more about what the next steps for this pediatrician should be!

Let's begin with some definitions. Dysmorphology is the study of causes, prognosis, treatment and prevention of congenital anomalies or basically the study of differences or alterations of typical morphology. Congenital anomalies are broad structural or functional defects present at birth. They may be internal (e.g., cardiac defects) or external (e.g., limb malformations). It is important to distinguish these from dysmorphic features, which are physical traits that are outside the range of typical human variation. These often involve the face, extremities, or body proportions and may be part of a larger syndrome. While dysmorphic features may have no

functional consequence, they can provide important clues to underlying genetic syndromes or undiagnosed congenital anomalies. Why is this important to know? Because according to the World Health Organization, congenital anomalies are a significant cause of infant morbidity and mortality worldwide (1). In Canada, government data shows that about 5% of infants with a congenital anomaly die in their first year of life, highlighting the need for early identification and referral (2). Among the most common congenital anomalies in Canada are undescended testes, congenital heart defects, and renal agenesis or hypoplasia. Once the healthcare provider can successfully identify anomalies, they are well on their way to helping a family determine the cause of the physical difference.

The origins of dysmorphic features lie in abnormal morphogenesis — the development of cells, tissues, and organs during embryonic growth. Perturbations in this process can manifest in different ways. A malformation sequence, such as a Pierre Robin sequence, is a single, localized issue with formation that initiates a chain of subsequent defects. A deformation sequence, such as some forms of clubfoot, is when an extrinsic intrauterine mechanical force, for example, uterine constraints, causes altered structural formation A disruption sequence, for example, a rubella infection, is when the fetus is exposed to a destructive problem and as a result has a vascular, infectious, or mechanical alteration. Dysplasia occurs when the primary defect is a lack of organization of cells into tissues; an example of this would be Marfan syndrome. Lastly, a malformation syndrome is when the patient has multiple structural defects in one or more tissues that are caused by a single initiating issue, for example an environmental teratogen, chromosomal abnormality or single gene disorder. An example of this is Trisomy 21 or Down syndrome. These malformations can be minor, meaning it is of no serious medical or cosmetic consequence to the patient, and does not require medical or surgical interventions. Or major, which is more concerning, potentially life altering, and requires significant interventions (3) If we consider a patient with Down Syndrome to help us understand these definitions better, a single palmar crease or low set ears would be minor malformations, and a congenital heart defect would be a major malformation.

When the clinician first encounters a child with dysmorphic features, there are key components in both a history and physical exam that should be considered. When completing a history, it is critical to inquire about the pregnancy, including exposure to teratogens such as maternal diabetes, alcohol, drugs (prescribed & non-prescribed), infections (fever, rash) and signs of a vascular disruption such as bleeding. In addition, gestational age at birth and mode of delivery are key questions. A detailed history should also ask about any prenatal ultrasound abnormalities and abnormal prenatal genetic screening results such as first trimester screening, maternal serum screening or non-invasive prenatal screening (NIPS). Asking about fetal movements is also helpful to identify the possibility of an underlying neurological issue. A neonatal history should be taken, including growth parameters such as birthweight, length, and head circumference. Ensure you ask about the need for resuscitation after delivery, NICU admission and any need for ventilatory support (such as CPAP or intubation), hypoglycemia or jaundice. Past medical history including any prior surgeries should be collected. Additional history should include a detailed review of systems looking for any feeding issues, seizures, abnormal tone, vision or hearing concerns, problems with elimination, birthmarks, unusual physical features, concerning behavioural patterns etc. A comprehensive family history is needed to assess for the possibility of inherited conditions, and a pedigree spanning 3 generations should be created. Questions should include ethnic background, consanguinity,

and other members of the family with similar problems, congenital anomalies, intellectual disability, developmental delay, autism, recurrent miscarriages, neonatal deaths or stillbirths.

With respect to the physical exam, it is important to follow a systematic approach to ensure a comprehensive and thorough assessment. The approach suggested here is from the Oxford Desk Reference for clinical genetics and genomics (4). Firstly, measure the growth parameters, and comment on the stature and build of the child or infant. Describe the shape of the cranium and size of the fontanelles, using words like large, small, or abnormal. Next move onto the face. When describing the facial features, look closely at the ears, eyes, nose, philtrum, mouth, palate, uvula and chin. For each feature, describe the size, angle/slope, and whether any features are distinctive or unusual. You can comment on the spacing of the eyes and whether there is hypertelorism or increased spacing between the eyes. And whether the palpebral fissures (opening of the eye) are horizontal, upward or downward slanting. Make your way down to the hands, feet, and limbs, noting any unusual nail findings, missing or extra fingers, or unusual palmar or plantar creases. Describe the chest shape, spacing of the nipples, back alignment, and perform a physical exam of the heart, abdomen, and nervous system. Ensure that the genitalia are examined. Comment on any skin changes including unusual pigmentation or birthmarks. Lastly, comment on the child's behaviours, mannerisms, stereotypies, and speech.

With the findings of the history and physical exam, a differential diagnosis can be generated. Organizing the differential diagnosis into broad genetic categories can help. The first is chromosomal abnormalities, with the more common conditions including trisomy 21, or Down syndrome, trisomy 18, or Edwards syndrome, trisomy 13, or Patau syndrome, and monosomy X, or Turner syndrome. These are also referred to as the common aneuploidies. The dysmorphic features of Down syndrome, given how common it is, are important to mention. Infants with Down syndrome can present with upward slanting palpebral fissures (or the opening of the eyes having an upward slant), hypertelorism (or widened spacing between the eyes), single palmar creases, hypotonia (or low muscle tone), and often have heart defects of the atrioventricular canal. More information on Down syndrome and Edwards syndrome can be found on the PedCases website. Next, consider some common microdeletions syndromes such a 22q11.2 deletion syndrome also known as DiGeorge syndrome, or a 7q11 deletion also known as Williams syndrome. You can also have microduplications syndromes. Next we have single gene disorders. These might include Noonan syndrome or Marfan syndrome. And lastly, it is important to keep in mind that some conditions with multiple congenital anomalies or dysmorphic features may be non-genetic including some of the teratogenic syndromes that can occur as a result of toxin exposures in pregnancy. These include fetal alcohol spectrum disorder, maternal diabetes, and maternal infections such as cytomegalovirus, toxoplasmosis, varicella or chicken pox, and rubella.

Next, we will discuss the work up of the child or infant with dysmorphic features and when to refer to a medical geneticist. First, consider the family history. Refer anyone with a family history of a genetic condition, birth defect, chromosomal disorder, or hereditary cancer. Make sure to inquire about ethnic background as well, because some genetic disorders are more common in certain groups. Next, refer a patient whose parents are blood related, who has had two or more pregnancy losses, a stillbirth, or has had a baby who died from unexplained causes. And finally, refer anyone who received results from third party testing and want to discuss implications of the results, or who has had any abnormal result suggesting a genetic condition (5). Initial

investigations can be undertaken by a pediatrician and include a rapid aneuploidy detection, or RAD, if a common aneuploidy is expected or a chromosomal microarray as a starting point. If a condition only affects one organ, a pediatric subspecialist might be appropriate to consult to order a gene panel. A medical geneticist can be consulted if additional more specialized investigations are needed including exome sequencing or there is a multisystem issue. If trisomy 21, 18, or 13 is expected, the patient DNA will be tested using karyotyping or RAD. A microarray test looks at deletions, duplications or copy number variants in the DNA. It is usually a blood test, and the results are available in 2-4 weeks. If this does not yield a result, the geneticist can order a gene panel to look at a target set of genes or cast a wider net and order a whole exome sequencing to investigate all protein-coding regions.

Other investigations to consider will be dependent on your physical exam findings. These investigations may include imaging to screen for congenital anomalies, such as an MRI of the brain, echocardiogram, renal ultrasound and skeletal survey. Other specialized investigations may include a hearing screen and formal eye exam. For example, if the child were suspected to have a skeletal dysplasia, a skeletal survey would provide useful information to help guide additional genetic investigations (6). If there is a suspicion for an inborn error of metabolism (ie a metabolic disorder), specialized biochemical genetic investigations may also be considered.

Now let's go back to Roger, the 6 month-old infant. We know that Roger has low muscle tone, failure to thrive, and a heart murmur. When speaking with the parents, you discover that Roger was born at full term and had an uncomplicated pregnancy and delivery. Roger did not stay in the NICU and has had no illnesses or hospitalizations so far. Roger's family history is unremarkable – there is no consanguinity between his parents, and there is no family history of any genetic conditions, congenital heart disease, or developmental disorders. You pull up his growth chart on Netcare and see that his weight and length on the typical WHO growth chart are both at the 5th percentile, with a head circumference at the 25th percentile. On examination, you notice some dysmorphic features. You see a broad forehead, bitemporal narrowing, periorbital fullness, a stellate or lacy iris pattern, a short nose with a bulbous nasal tip, a wide mouth, full lips, mild micrognathia, epicanthal folds, full cheeks, and a flat facial profile (7). You confirm the presence of a systolic murmur heard in the left upper sternal border and notice some mild hyperextensible joint with no organomegaly. Given what we have learned so far, what would your next steps be? First, you order an ECG and echocardiogram to investigate the heart murmur and it confirms the presence of a supravalvular aortic stenosis. You decide to get a microarray done to test for the presence of a 7q11 deletion, seen in Williams Syndrome, which comes back positive. Good job! You have successfully diagnosed a genetic condition in this patient and can now refer him to the relevant specialists including cardiology and genetics who will counsel the family on next steps.

To summarize what we have learned so far, approaching a child with dysmorphic features requires a detailed prenatal, neonatal, medical, developmental and family history, a structured physical exam focused on subtle patterns, a thoughtful differential diagnosis using categories, and lastly, early referral to genetics and appropriate investigations. Recognizing dysmorphic features is a critical clinical skill that often leads to earlier diagnosis, better outcomes, and informed family planning. We hope this approach to the child with dysmorphic features has been helpful in developing a comprehensive and systematic approach to recognizing and investigating those who fall into this population. References and a podcast script are available on the PedCases website. Thanks for listening!

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