Divine Intervention Episode 17 Diseases of The Pediatric Population Part 1

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Some MS4

1

7 yo Asian M with chronic watery diarrhea and bloating. Stool is +ve for reducing sugars. A hydrogen breath test is +ve. Diagnosis? Treatment?

2

23 mo is brought to the pediatrician by his mom. PE is notable for pronounced bowing in the LEs. MCV is 60 and serum Ca is 7.1. He has an erythematous, blistering rash in front of both patellae. There is diffuse muscle wasting. Diagnosis? Dx testing? Treatment? Anaphylactic reaction to transfusions?

3

8 mo that consumes cow milk is brought to the pediatrician by his mom after noticing blood in his stool last week. CBC is notable for anemia. Diagnosis? Treatment?

4

5 yo kid with chronic constipation. Immigrated from S. America at the age of 1. A rectal exam is accompanied by an explosive expulsion of poop. Distal colon biopsy reveals an absence of Auerbach's plexus. Diagnosis? Other patient populations? Treatment?

5

18 mo with a recent history of an Upper Respiratory infection is brought to the ED by his mom. He has been having “paroxysms” of abdominal pain that have made him “curl into a ball”. His mom brings his most recent diapers to the office with some of them having a red tinge. A mass is palpated in the RLQ of the abdomen. Diagnosis? Diagnostic testing? Treatment? Pathophysiology?

6

4 yo presents with severe, generalized abdominal pain, nausea, and vomiting that started 12 hrs ago. CBC is notable for a WBC of 13k (high). US reveals an echogenic material in the RLQ with what appears to be fat stranding. Diagnosis? Treatment? Complications?

7-The Classically Tested Genetic Principle

Myotonic dystrophy/Fragile X Syndrome. Angelman/Prader-Willi Syndrome/Beckwith Wiedemann Syndrome (2). Trisomy 21. Mom passing a disease to all her kids. Quick aside on the VACTERL association.

8-Downs Syndrome

Genetic pathophysiology. Early onset neuro dz. Classic hematologic malignancy. Considerations before beginning sports. Hand findings. Common cardiac defect. Common GI defects (2). Maternal quad screen findings. How many chromosomes are found in a “Robertsonian Translocation” (RT) carrier? How many chromosomes are found in a patient with Downs Syndrome from a RT?

9-What is your diagnosis?

Prominent occiput + overlapping digits + clenched fists + rocker bottom feet.

Vs

Rocker bottom feet + Cleft lip/palate + Polydactyly.

10

5 yo M with a long face, large ears, and large testicles. Diagnosis? Mechanism of inheritance? Genetic pathophysiology (+ associated principle)? Diagnostic testing? Common psychiatric association? Common neuro association?

11

Tall male + Gynecomastia + Infertility + No facial hair. Diagnosis? Karyotype?

12

Female with a low posterior hairline, webbed neck, congenital lymphedema, and short stature. Diagnosis? Karyotype? Aortic pathologies (2)? Renal pathology? How can the short stature be corrected? How can the development of secondary sexual characteristics be promoted at puberty? What is the “kind” of hypogonadism associated with this disorder? As an aside, what is the male equivalent of Turner’s syndrome? Is this disorder only present in males? What is the mechanism of inheritance? Genetic mutation? What is one good “cardiac clue” on exams to help with differentiating this counterpart from Turner’s syndrome?

13

CATCH-22 syndrome. The 2 mechanisms behind the development of Prader Willi syndrome. The 2 mechanisms behind the development of Angelman syndrome. Classic presentation of both disorders. The CHARGE association.