GENETIC AND METABOLIC DISORDERS

Case 1: 15 yo. ♀ w/ genovarum, no teeth, short stature

→ hand x ray showed typical features of rickets (cupping, widening, fraying, ↓ bone density)

→ bone profile (calcium, phosphate, alkaline phosphatase)
normal ↓ ↑

∴ hypophosphatemic rickets
(X linked dominant, most common inherited rickets)

Case 2: 5 mo. old, lethargic, distended abdomen (massive hepatomegaly)

→ check glucose levels → 20 → give glucose → 🙂

∴ glycogen storage disease
(most types are autosomal recessive)

features: full cheeks, more hair (longer lashes, ...)

* pts w/ chronic diseases usually have higher androgen sensitivity → more hair growth

* hypoglycemia + microgenitalia → hypopituitarism (↓ GH)
(not glycogen storage D)

Case 3: hypoglycemia + macroglossia + facial hemangioma + umbilical hernia

→ do critical sampling of insulin (measure at the peak of hypoglycemia) → high insulin
→ macrosomia + no ketones in urine + ear creases

∴ hyperinsulinemia (exogenous or endogenous)
Beckwith - Wiedemann Syndrome

Case 4: 6 mo. old, Chronic diarrhea, alopecia, dermatitis (severe napkin dermatitis)

∴ acrodermatitis enteropathica
(zinc deficiency, autosomal recessive)
→ give zinc

Case 5: 10 yrs, severe abdominal pain & tenderness, skin lesions on elbows & knees (orange!)

→ do lipid profile

∴ hyperlipidemia → congenital acute pancreatitis (autosomal dominant)

Case 6: diagnosed w/ gallbladder stone
+ he has photo dermatitis (only on sun exposed areas) + dark teeth + blisters + paronychia

∴ erythropoietic porphyria (autosomal dominant)
a type of cutanous porphyria

Case 7: Chronic fever, full lips, light eyebrows, teeth problem

hair, nails, teeth, sweat glands → ectoderm

(reason of fever is the absence of sweat glands as sweat reduces temp.)

∴ ♂ anhydrotic ectodermal dysplasia (X linked)
♀ hypohidrotic ectodermal dysplasia (AD)

Case 8: Short stature + severe distension (massive hepatosplenomegaly) + pancytopenia

∴ lipid storage disease (Gaucher's disease)
→ require enzyme replacement (every 2 wks for life)

→ if enzyme therapy → do splenectomy as a temporary measure to reduce distension (pt will mostly die in 2 yrs)

Case 9: pt w/ ataxia & telangectasia

∴ Ataxia-telangectasia syndrome (autosomal recessive, defect in DNA repair → more prone to malignancy)

pt died 2 years later of lymphoma

Case 10: Unilateral increased tearing (epiphoria) + proptosis + photophobia (triad)

∴ congenital glaucoma
early diagnosis saves vision

→ presentation in neonates: macrocornia

Case 11: heterochromia iridis + deafness + white strand of hair

∴ Waardenburg syndrome (autosomal dominant)

Case 12: blue sclera + fractures

∴ osteogenesis imperfecta (↓ collagen)

Case 13: brown spots on trunk (café au lait spots)
axillary freckling
scoliosis

∴ Neurofibromatosis (autosomal dominant)
(plexiform neurofibroma)
a type of neuro cutanous syndromes

Case 14: facial lesions (look like acne), convulsions,
hypomelanoic patches (at least 3),
Shagreen patches

∴ tuberous sclerosis
a type of neurocutanous syndromes

Case 15: lt sided hemiplegia / rt sided port wine stain

∴ sturge weber syndrome (sporadic)
rt MCA occlusion caused lt hemiplegia
may also be associated with ipsilateral
glaucoma, seizures, developmental delay

* middle meningeal artery → epidural hematoma

Case 16: excessive hair growth, cloudy eyes, short stature,
hepatosplenomegaly, everted umbilicus

∴ hurler syndrome (autosomal recessive)
a type of mucopolysaccharidosis

* hunter is another type (x linked)

Case 17: 11 yo. ♂, severe cachexia, finger sucking, unable
to walk, still uses diapers
urine smell like maple syrup (unable to digest AAs
valine, leucine, isoleucine)

∴ maple syrup urine D (need special milk formula)

Case 18: developmental delay, musty smell of urine, blonde, blue eyes

∴ PKU (need special milk formula)

Case 19: Down's syndrome, bile stained vomiting, double bubble sign

∴ Congenital duodenal obstruction

Case 20: tall ♀, visual problem, arachnodactyly

∴ marfan syndrome (autosomal dominant)

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* raccoon eyes, CSF leak → basal skull #

* most common cause of inherited mental retardation: fragile X syndrome.

* dorsal edema:
  ♀ → turner syndrome (may be with coarctation or bicuspid aortic valve → lt sided heart disease)
  ♂ → noonan syndrome (mostly rt sided heart disease)

* most common cause of inherited short stature: achondroplasia (autosomal dominant)

* thumb abnormality + anemia → fanconi syndrome

* extreme obesity → uniparental disomy

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