Mental Retardation

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Lecture Organization

- Mental Retardation Introduction
- Diagnostic Two Step
- Review of Individual MR Etiologies
  - Down's Syndrome
  - Edward's Syndrome
  - Fragile X Syndrome
  - Lesch-Nyhan
  - Phenylketonuria
  - Prader-Willi
  - Angelman Syndrome
  - Williams Syndrome
  - Fetal Alcohol Syndrome
  - Cri Du Chat Syndrome

2

Question: A child born to a 43 year old mother is diagnosed with Down Syndrome. On the second day of life massive emesis begins. What is the infant's most likely medical complication?

A. Atrial Septal Defect
B. Congenital Hypothyroidism
C. Duodenal Atresia
D. Hypotonia of gastro-esophageal sphincter
E. Macroglossia

3

Question: FISH (Fluorescent In Situ Hybridization) can be used to diagnose all of the following except?

A. Angelman Syndrome
B. Autism
C. Down Syndrome
D. Prader-Willi Syndrome
E. Williams Syndrome

4

Question: A child presents with mental retardation, eczema, repeated vomiting and seizures. The mother was told that she must have missed post-natal screening. The infant is most likely to have which of the following?

A. Fragile X Syndrome
B. Lesch-Nyhan Syndrome
C. Phenylketonuria
D. Rubella
E. Tay Sachs Disease

5

Question: Which of the following is the most common inherited cause of mental retardation?

A) Down's Syndrome
B) Edward's Syndrome
C) Fetal Alcohol Syndrome
D) Fragile X Syndrome
E) Wilson Disease

6

Question: A mother brings in her 7 year old son, whose IQ is 65. The mother reports the child's history of self injurious behaviors, a recent diagnosis of renal failure, severe gout, and poor muscle control. What is the most likely metabolic deficiency associated with this syndrome?

A. Phenylalanine Hydroxylase in Phenylketonuria
B. Dopamine Hydroxylase deficiency
C. Alkaline Phosphatase in Gushers disease
D. Purine metabolism in Lesch-Nyhan Syndrome
E. Ceruloplasmin in Wilson disease

7

Question: Which testing instrument is most applicable to test the intellectual functioning in a 4 year old child with hearing impairment?

A. Leiter-R International Performance Scale
B. Peabody Picture Vocabulary Test
C. Stanford–Binet Intelligence Test
D. Vineland Adaptive Behavior Scale
E. Wechsler Preschool and Primary Scale of Intelligence (WPPSI)

8

Question: A 10 year old mentally retarded male is on medication for ADHD. He has a long face, prominent large ears, hyper-flexible joints and macroorchidism. What is the likely diagnosis?

A. Fragile X Syndrome
B. Lesch-Nyhan Syndrome
C. Prader-Willi Syndrome
D. Turner's Syndrome
E. Williams Syndrome

9

Question: The Individuals with Disability Education Act (IDEA) was passed to accomplish which of the following?

A. Allow mentally retarded individuals to live in group homes and to receive education
B. Mandates free appropriate public education in least restrictive environments
C. Allow the mentally retarded to apply for social security
D. Allow the mentally retarded to decide which parent will obtain guardianship in case of parental divorce

10

Question: A newborn with an APGAR score of 9 undergoes standard testing for phenylketonuria (PKU) 12 hours after birth. The parents are relieved that the results are negative. The appropriate interpretation of these results is which of the following?

A. The negative results conclusively rule out the presence of PKU.
B. The test for PKU is valid only after the newborn ingests a diet containing phenylalanine.
C. The test for PKU has low sensitivity and standard protocol is to repeat it three times.
D. The PKU test is not done routinely in newborns in the US. It is done only when APGAR scores are below 7.

11

Mental Retardation: Overview

- IQ=Mental age/ Chronological age X 100
  - IQ remains stable throughout life
  - Mean IQ is 100, with Standard Deviation of 15
  - Average IQ range: 90-110
  - IQ of 70-84 classified as Borderline IQ
- Epidemiology of MR
  - 1% of population with MR
  - 35% genetic cause found
  - 10% with a malformation syndrome present

12

Mental Retardation: DSM-IV Diagnosis

1. Significantly subaverage intellectual functioning – IQ below 70
2. Impaired adaptive functioning
3. Onset before age 18

13

Mental Retardation: Levels

- **Borderline Intellectual Function**
  - IQ 70- 84 – V Code
- **Mild MR**: IQ 50-55 to 70 (85% of total)
  - Academic skills equal to 6th grade
  - Adults achieve minimal self-support and employment but need supervision
- **Moderate MR**: IQ 35-40 to 50-55 (10% of total)
  - Academic skills equal to 2nd grade
  - Live in supervised settings, perform unskilled work
  - Can bathe and prepare simple meals

14

Mental Retardation: Levels

- **Severe MR**: IQ 20-25 to 35-40 (3-4% of total)
  - May be taught to sight read "survival words"
- **Profound MR**: IQ below 20-25 (1-2% of total)
  - Impaired sensorimotor function, highly supervised
- **Mental Retardation, Unspecified**
  - Strong presumption of MR but untestable

15

Areas Of Adaptive Function: DSM-IV

- Communication
- Self-care
- Home living
- Social / interpersonal skills
- Use of community resources
- Self-direction
- Functional academic skills
- Work
- Leisure
- Health
- Safety

16

MR: Areas Of Disturbance

- MR: Global disturbed brain development
  - Intellectual disturbances
  - But also behavioral / psychiatric disturbances
- Intellectual Impairment
  - Attention, memory, language, praxis, reasoning
- Psychiatric & Behavioral Dysfunction
  - Depression, mania, anxiety, psychosis, eating, sexual, sleep and impulse disturbances
  - Self-injurious behavior, violence, agitation
- Concept of "Behavioral Phenotype"

17

Mental Retardation: Diagnostic Two Step

- Step 1: Diagnose the MR Syndrome
  - Assess intellectual function: IQ Test
  - Assess Adaptive Function: assessment interview(s) & rating instruments (Vineland)
  - Assess for behavioral / psychiatric disturbances

- Step 2: Diagnose the MR Etiology
  - Complete medical work up

18

Step 1: Assess MR Syndrome

- Step 1a: Assess intelligence
  - Bayley Scale for Infant Development
  - WAIS, WISC, WIPSSI
  - Leiter International Performance Scale
- Step 1b: Assess adaptive skills
  - Vineland Adaptive Behavior Scale
- Step 1c: Assess psychiatric and behavioral manifestations / comorbidities of MR

19

Bayley Scale For Infant & Toddler Development

- What: test of development
- Who: children 1 - 42 months who may be at risk of developmental delay
- Details: Components
  - Cognitive Scale: sensory, memory, learning, problem-solving skills
  - Language Scale: receptive and expressive
  - Motor Scale: fine and gross motor skills
  - Social-Emotional & Adaptive Behaviors Caregiver Questionnaire

20

Bayley Scale Instructions To Parent

"I will be giving CHILD some toys and watching what (he/she) does with them. Sometimes I will ask (him/her) to do specific things with the toys. For example, I will ask (him/her) to put cubes in a cup, to put some pegs in a board, and to name some objects. Some of the things I'll ask will be easy, some will be too hard, and some will be just right. We use this range of tasks so we can find out all the things CHILD is doing right now. While we're doing this, please try not to talk to CHILD because I want (him/her) to pay attention to me."

21

Wechsler Intelligence Scales

- Wechsler Preschool and Primary Scale of Intelligence (WPPSI) – For ages 2y 6m to 7y 3m
- 14 subtests that can generate the following scores
  - Verbal IQ
  - Performance IQ
  - Full scale IQ
  - General language
  - Processing speed

Vocabulary assessment through picture naming

22

Wechsler Intelligence Scales

- Wechsler Intelligence Scale for Children (WISC-IV) – For ages 6 -17
- 10 core subtests grouped into 4 indices
  - Verbal comprehension
  - Perceptual reasoning
  - Working memory
  - Processing speed

23

Non-Verbal IQ measures

- Used for children with communication disorders or hearing impairment
- Leiter International Performance Scale Revised (Leiter-R)
- Test of Nonverbal Intelligance-3 (Toni-3)
- Comprehensive Test of Nonverbal Intelligence (CTONI) (computerized test)
- Peabody Picture Vocabulary Test

24

Leiter International Performance Scale

- What: Test non-verbal intelligence (No need for spoken language, not even for instructions)
- Who: cognitively delayed, disadvantaged, non-English speaking, hearing impaired, speech impaired, or autistic persons 2-20 yrs
- Details: 4 subtests
  - Reasoning
  - Visualization
  - Memory
  - Attention

25

Vineland Adaptive Behavior Scale

- What: Measure of Self-Sufficiency
- Who: Intellectual, developmental delay, autism spectrum, ADHD
- Details: 2 Formats: 1) Semi-structured interview with care-giver and 2) survey report of teacher
  - Communication: receptive, expressive, written
  - Socialization: play, leisure, coping, and interpersonal
  - Daily Living Skills: personal, domestic, community
  - Motor Skills: gross, fine
  - Maladaptive Behaviors: Internalizing, Externalizing

26

MR: Psychiatric Differential Diagnosis

- Pervasive Developmental Disorders
- Learning Disorders
- Communication Disorders
- Borderline Intellectual Functioning

27

MR: Comorbid Diagnoses

- Prevalence of psychiatric disorders
  - 30-70% of MR individuals
  - 4-6 x that of general population
- Diagnosing Comorbidities
  - Referrals to psychiatrists likely not for MR but for emotional/behavioral problems
  - Focus on change from baseline and objective symptoms rather than on subjective symptoms
  - Take more time and an informal interview style
  - Always consider trauma/abuse in differential (due to vulnerability in MR population)

28

MR: Highly Comorbid Disorders

- Pervasive Developmental Disorders
- ADHD: similar to rates in non-MR
- Pica, Rumination, Tic Disorders
- Mood: depression and bipolar disorders
- Anxiety: OCD, PTSD
- Behavioral Dyscontrol
  - Self-injurious behaviors (head banging, self-biting): Lesch-Nyhan, Fragile X, Autism
  - Aggression: consider psychosis, depression, physical or sexual abuse

29

Specific Diagnoses Of Comorbidity

- MR from Fragile X: ADHD in 80%
- MR from Prader-Willi: OCD, depression
- MR from Williams: ADHD, anxiety, and depression
- MR from Down's: Alzheimer's dementia often after age 40 and depression

30

Diagnostic Step 2: MR Etiologies

- Genetic (Hereditary) abnormalities
  - Tay-Sachs, Tuberous Sclerosis, Fragile X
- Chromosomal abnormalities
  - Down's (trisomy 21), Triple X, Turner's
- Abnormalities of embryonic development
  - Maternal infections, intrauterine toxin or alcohol exposure
- Pregnancy Problems
  - Fetal malnutrition, hypoxia, birth trauma
- GMCs
  - Childhood infections, trauma, toxins (lead)
- Environmental Factors (next slide)

31

MR: Environmental Factors

- Deprivation of nurturance
- Deprivation of social and linguistic exposure
- Abuse
- Social Economic Status (SES)
  - Mild MR more common in lower SES
  - Moderate, severe, and profound equal in all SES
- Mild MR: Complex Causation
  - Combination of polygenic inheritance and environmental factors such as prenatal difficulties, obstetrical problems, nutrition, and psychosocial nurturance

32

MR Etiologies: Review Of Concepts

- **Genetic Abnormalities**
  - Causal Genetic Abnormalities of specific genes
    - Inherited mutations
    - Sporadic mutations
  - Non-Causal Susceptibility Loci / Multigenetic Disorders
    - Combined effect of multiple loci (genes) each of which makes a weak contribution to disorder
- **Chromosomal Abnormalities**
  - Often not inherited - they occur during meiosis
  - Crossing-Over, Translocations and Trisomies

33

Chromosomal Abnormalities

- Diploid Genome: 46 chromosomes
  - 22 pairs of autosomes and 1 pair sex chromosomes
  - Meiosis: cell division in reproductive organs, resulting in gametes containing 23 chromosomes
- Translocations: chromosomes break and fragments combine with other chromosomes
  - Unbalanced translocations: unequal translocated fragments – Familial Down's Syndrome (2-3%)
- Trisomies: during meiosis, chromosomes may fail to separate (nondisjunction) and these gametes end up with a trisomy and monosomy

34

Chromosome Lingo

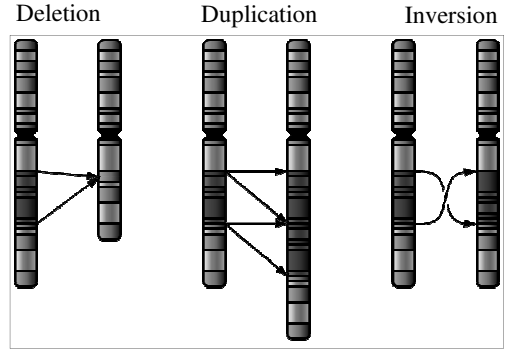
p arm (for "petite")

q arm (because q follows p in alphabet)

Example: 18p11.13 means 11.13 region of short arm of chromosome 18

35

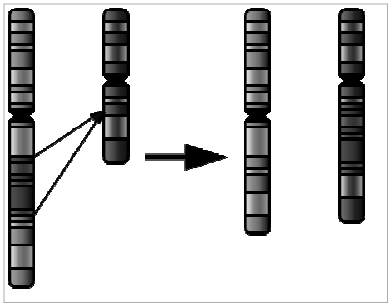
Single Chromosome Mutations



36

Two Chromosome Mutations

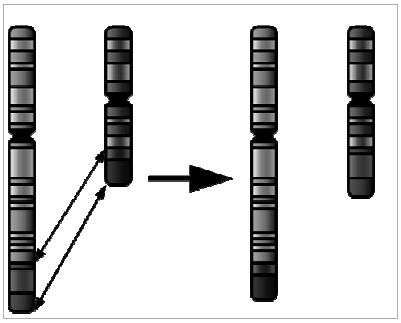
Insertion



37 Before After

Two Chromosome Mutations

Translocation



38 Before After

Step 2: Assess For Etiology

- Assess for Environmental Contributors
  - Neglect, deprivation, abuse, nutritional deficits
- Assess for Medical Etiologies
  - PE: dysmorphisms, hearing and vision tests
  - Metabolic eval: serum organic acids, lactate, pyruvate, TSH, triglycerides, carnitine, lead level
  - Urine: mucopolysaccharides, amino acids
  - MRI
  - Genetic Analysis: esp. with dysmorphisms

39 Adapted from AACAP Parameters

Fluorescent In Situ Hybridization

- FISH: A test for identifying chromosomal and genetic abnormalities
  - Downs, Angelman, Prader-Willi, Cri-du-Chat, Velocardiofacial Syndrome
- How It Works
  - Identifies and localizes DNA sequences
  - Probe: a sequence of DNA tagged with fluorophores
  - Hybridization: probe attaches to single strand DNA from patient. Fluorescence seen under microscope

40

Tests Of Newborns: Most Common

- Phenylketonuria (PKU)
- Congenital Hypothyroidism
- Galactosemia
- Maple Syrup Urine Disease
- Homocystinuria
- Biotinidase
- Sickle Cell Disease
- Congenital Adrenal Hyperplasia
- Cystic Fibrosis

41

Mental Retardation: Most Common Etiologies

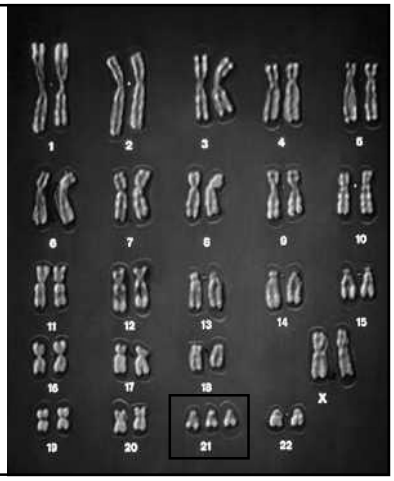
- Top 3 causes of MR account for 30% of cases:
  - Down's Syndrome (trisomy 21): most common genetic (or chromosomal) cause
  - Fragile X syndrome (X Chromosome gene FMR-1): most common inherited cause
  - Fetal Alcohol Syndrome: most common preventable cause

42

Down's Syndrome: Trisomy 21

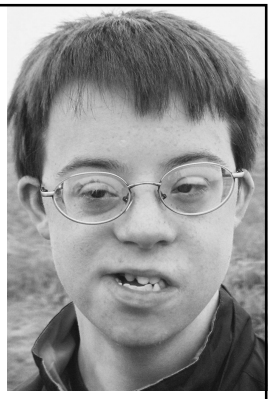
- Trisomy 21 Down Syndrome
  - Abnormal gamete + normal gamete
- Mosaic Down Syndrome
  - 2 normal gametes
  - Nondisjunction occurs during early cell division
  - Some cells normal, others trisomic

43



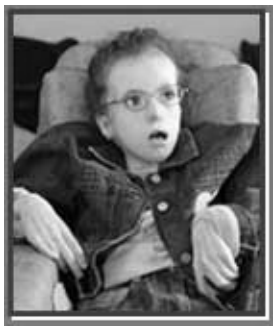
Down's Syndrome Features

- Shortened palpebral fissures
- Macroglossia
- Iris white spots (Brushfield spots)
- Deep palmar transverse crease
- Hypotonia, incoordination
- Congenital hypothyroidism
- GI Atresias
- Atrial / ventricular septal defects
- Alzheimer's Dementia at 40



44

Edward's Syndrome: Trisomy 18



- Meiotic nondisjunction
- Survival
  - 95% die in utero
  - Of live born infants only 5-10% one year
- Features
  - Severe MR
  - Severe renal / cardiac / intestinal malformations

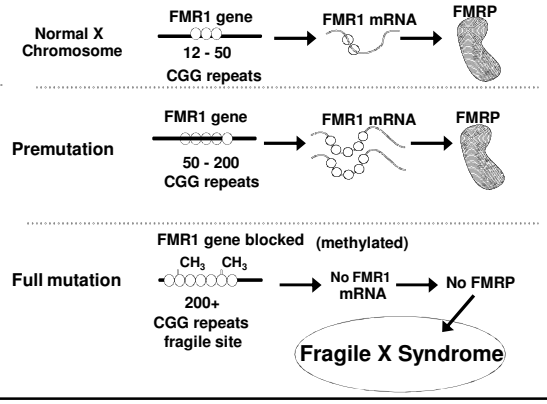
45

Fragile X Syndrome

- Family of genetic mutations that increase the number of CGG repeats on the long arm of the X chromosome
- These CGG repeats disable the Fragile X Mental Retardation 1 gene (FMR-1 gene) by causing methylation of the regulatory region of this gene.
- The disabling of the FMR-1 gene leads to lack of Fragile X Mental Retardation Protein (FMRP) which, in turn, results in clinical presentation.

46

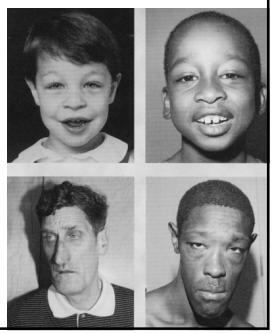
Fragile X Premutation & Mutation



47

Fragile X Syndrome Features

- Long face, large ears, prominent jaw, soft skin, macroorchidism, mitral valve prolapse
- MR, learning disorders, speech problems
- Hyperactivity, ADHD,
- Autism in about 33%



48

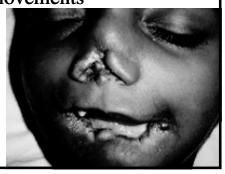
Other Fragile X Related Syndromes

- Fragile X Associated Tremor / Ataxia Syndrome (FXTAS)
  - Present in premutation carriers – ages 50-80
  - Intention tremor, ataxia, affective instability and personality changes
- Fragile X Associated Premature Ovarian Insufficiency (FXPOI)
  - Defined as menopause < age 40
  - ~ ¼ FMR1 premutation carriers get FXPOI
  - ~ ¼ get premature menopause defined as < age 45

49

Lesch-Nyhan Syndrome

- Deficiency in Purine Metabolism
  - Hypoxanthine-guanine-phosphoribosyl transferase (HPRT): increases uric acid
- Features
  - Severe MR
  - Self injury - lip, finger biting, scratching
  - Severe gout, poor muscle control, joint and renal malformations
  - Grimacing, writhing and repetitive movements
- Sequelae
  - Deficiency in vitamin B12
  - Basal Ganglia damage
  - Renal failure
  - Death



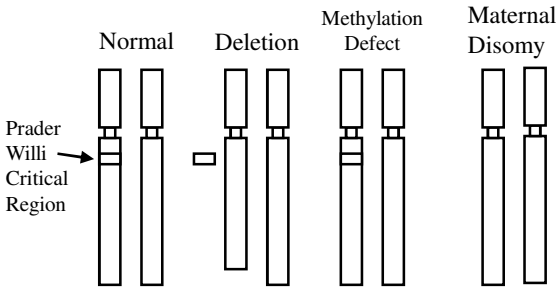
50

Chromosome 15 Abnormalities: Prader Willi And Angelman

- Maternal and paternal homologue chromosomes 15 express certain genes differently
  - Due to sex-related epigenetic imprinting
  - The biochemical mechanism is DNA methylation
- When maternal genes are deleted or inactivated, the result is Angelman Syndrome
- When paternal genes are deleted or inactivated result is Prader-Willi Syndrome

51

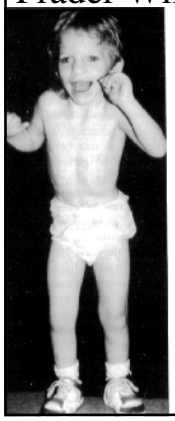
Prader Willi Genetic Causes



Paternal genes deleted, not activated, or not present

52

Prader Willi: Features




- Mental retardation
- Small hands / feet
- Morbid obesity
- Almond eyes
- Scoliosis
- Undescended testicles
- Hypotonia

Prader Willi: Psychiatric Features

- Most with IQ between borderline and mild Mental Retardation
- Obsessive Compulsive symptoms
- Insatiable appetite
- Agitation if denied food
- Frequent skin picking

54

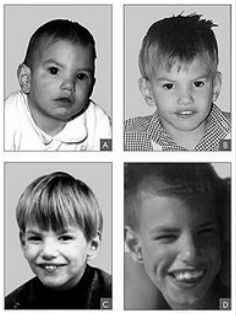
Angelman Syndrome

- Deletion / inactivation of part of maternal chromosome 15
- Intellectual / developmental delay, seizures, hand flapping, sleep disturbance, happy / smiling demeanor
- "Angels" – young and happy
- Epidemiology: 1 in 20,000

55

Cri-du-Chat Syndrome

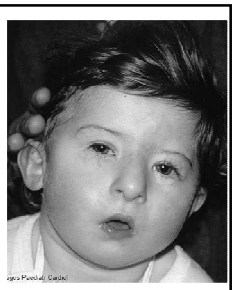
- Chromosome 5p Deletion
  - 90% sporadic 5p deletion: monosomy
  - 10% 5p translocation: monosomy + trisomy of another part of genome
- Features
  - Severe MR
  - Mewing cat sound
  - Hypotonia, microcephaly, cardiac defects
  - Hyperactivity, aggression, repetitive behaviors
- Epidemiology: 1 in 20k -50k births



8m 2yrs
4yrs 9yrs

56

Velocardiofacial Syndrome



- Genetic microdeletion: 22q11
- Epidemiology:
  - 1 in 700 births
  - ≥5% of infants with cleft palate
- Features
  - Cleft palate (velum = palate)
  - Congenital cardiac malformations
  - Long face, almond-shaped eyes, wide nose
  - Hypoparathyroidism – hypocalcemia may lead to seizures
  - MR or learning disorders
  - ADHD

57

Williams Syndrome

- Genetic microdeletion 7q11.23
- Features
  - Mental Retardation
  - Elfin appearance
  - Diabetes mellitus
  - Supravalvular Aortic Stenosis
  - Hypercalcemia
  - Hearing loss (sensorimotor)
- Epidemiology: 1 in 7,500

58

Williams Syndrome: Features

- MR Features
  - Visuospatial deficits
  - Verbal and verbal memory relative strengths
  - Mild to moderate MR
- Psychiatric Features
  - Overfriendly personality – "cocktail party type"
  - Poor judgment – will befriend strangers
  - Anxiety Disorders: esp. specific phobia[1]
  - ADHD[1]
  - Depressive Disorders[1]

1. Kennedy et al, Jefferson JP, 2006

59

Smith-Magenis Syndrome

- Deletion of chromosome 17 region - loss of RAI1 Gene
- Features
  - Full downturned lips
  - Broad lower face / jaw
  - Flattened bridge of nose
  - Short, with scoliosis
  - Kidney & cardiac abnormalities
  - Reduced sensitivity to pain / temp
- Epidemiology: 1 in 15,000

60

Smith-Magenis Syndrome: Behavioral Features

- Sleep Disturbance
  - Sleep reversal – often up at night
- Hand clasping, self-hugging, finger-licking
- Self-injurious behaviors
  - Skin-picking, nail pulling, head banging
- Agitation
  - Aggression, temper tantrums, anxiety
- ADHD

61

Phenylketonuria

- Autosomal Recessive genetic disorder
  - Diagnosed by neonatal screening 2-7 days after birth
  - Deficient hepatic enzyme Phenylalanine Hydroxylase
  - It converts Phenylalanine to Tyrosine
  - Toxic phenylalanine & phenylketone levels
- Features
  - Severe mental retardation
  - Seizures in about 1/3rd of pts
  - Also, Eczema, vomiting
- Epidemiology: 1 in 1/10,000 births
- Course: Diet low in phenylalanine improves behavior and developmental progress

62

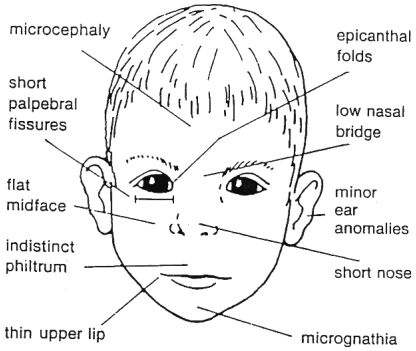
Fetal Alcohol Syndrome (FAS)

- Severe form of Fetal Alcohol Spectrum Disorders (FASD) - when some but not all FAS features present
- FAS most common preventable cause of MR
  - 1 in 750 to 1 in 5000 births (depending on region)
  - FASDs three times as common as FAS
- Features
  - MR or learning disorders; Physical and psychiatric sequelae
- Management
  - Substance Abuse Treatment for pregnant woman
  - Target also father to support mother
  - Target prevention efforts towards women who abuse substances and can become pregnant

63

Fetal Alcohol Syndrome Features

Also
- Incoordination
- Small stature
- Sleep and sucking abnormalities



64

FAS: Secondary Conditions

- Psychiatric
  - ADHD, Conduct Disorder, Substance Dependence, Depression, even Psychosis
  - Frustration, aggression, and impulse dyscontrol
- Life Problems
  - More likely to be suspended, expelled, drop out
  - Much higher rates of arrest / conviction
  - Promiscuity and inappropriate sexual behavior
  - Work problems
  - Parenting problems

65

Lead Poisoning

- GI: diarrhea, constipation, cramps, aches, nausea, vomiting, loss of appetite
- Cognitive: lower IQ, ADHD, irritability, aggression, lack of interest, changes in mood / personality
- General: headaches, fatigue, muscle weakness, sleep disturbance
- Risk <u>for</u> lead poisoning: Abuse / Neglect
  - 2/3 of neglected kids may have lead > 10 µg/dl[1]
  - Test all kids with abuse / neglect hx for lead

66

1. Flaherty, Clin Peds, 1995

HIV In Children

- Epidemiology: 9300 children under age 13
  - Almost all transmitted from mother: intrauterine and through breast-feeding
  - 25% of HIV+ mothers not on CART, transmit HIV to child
- Diagnosis
  - Use Polymerase Chain Reaction: must identify virus and not antibody. Maternal antibody persists for 18 m.
- Signs
  - Fast / Slow Progression: 20% / 80%
  - Microcephaly, developmental delay, ADHD, seizures
  - Motor: tremor, rigidity, dystonia, dyskinesia, ataxia

67

MR: Management Principles

- Early intervention to decrease MR morbidity
  - Normalization and community care - Special classes / programs in community schools
  - Caretakers to be active treatment planning participants
- Individuals with Disability Education Act (IDEA)
  - Entitles children to full range of diagnostic, educational and support services from birth to age 21
- Biomedical and Psychiatric Treatment
  - Assess and treat underlying conditions, and medical and psychiatric sequelae

68

MR: Treatment Of Psychiatric / Behavioral Conditions

- Behavioral intervention often first line
  - Training of patient, caretakers, teachers
- Medication Guidelines / Precautions
  - Must conduct comprehensive assessment prior to starting meds (except in emergency)
  - Establish clear goals of treatment including target s/s
  - Risk / Benefit: med may help target symptom but hurt in other areas of function
  - Obtain informed consent
  - Caution: anti-cholinergics & benzodiazepines

69

MR: Medications For "Disruptive Behaviors"

- Stimulants: methylphenidate, amphetamine
- Atypical Antipsychotics: risperidone
- Antidepressants: SSRIs
- Anticonvulsants: Valproate, Carbamazepine
- Adrenergics: clonidine, guanfacine
- Opioid Antagonist: naltrexone for self-injurious behaviors (mixed results)

70

MR: Behavioral Emergencies

- Ensure safety of patient and others
  - Redirect
  - May need to use physical restraints
  - Administer emergency medication
- Diagnose Cause
  - Change in medical status: illness
  - Thirsty, hungry, overstimulated, change in personnel or routine
  - Abuse: physical, sexual, psychological
- Implement Changes

71

Individuals With Disabilities Education Act (IDEA)

- Law that ensures educational access to 6.5 million eligible children
- Covers early intervention, special education and related services
- Mandates free appropriate public education in least restrictive environments

72

www.idea.ed.gov

MR: Course And Prognosis

- Development often slow but not deviant
  - There is a 'ceiling' on development, but proper training can improve function and thus prognosis
- The majority of MR (85%) is mild (IQ 50-70)
  - Can learn academic skills up to 6th grade level,
  - Are flexible in adapting to environment and need minimum self support
- Complications arise from delayed speech, behavioral problems & comorbidities, societal attitudes, financial limitations

73

Question: A child born to a 43 year old mother is diagnosed with Down Syndrome. On the second day of life massive emesis begins. What is the infant's most likely medical complication?

A. Atrial Septal Defect
B. Congenital Hypothyroidism
C. Duodenal Atresia
D. Hypotonia of gastro-esophageal sphincter
E. Macroglossia

74

Question: FISH (Fluorescent In Situ Hybridization) can be used to diagnose all of the following except?

A. Angelman Syndrome
B. Autism
C. Down Syndrome
D. Prader-Willi Syndrome
E. Williams Syndrome

75

Question: A child presents with mental retardation, eczema, repeated vomiting and seizures. The mother was told that she must have missed post-natal screening. The infant is most likely to have which of the following?

A. Fragile X Syndrome
B. Lesch-Nyhan Syndrome
C. Phenylketonuria
D. Rubella
E. Tay Sachs Disease

76

Question: Which of the following is the most common inherited cause of mental retardation?

A. Down's Syndrome
B. Edward's Syndrome
C. Fetal Alcohol Syndrome
D. Fragile X Syndrome
E. Wilson Disease

77

Question: A mother brings in her 7 year old son, whose IQ is 65. The mother reports the child's history of self injurious behaviors, a recent diagnosis of renal failure, severe gout, and poor muscle control. What is the most likely metabolic deficiency associated with this syndrome?

A. Phenylalanine Hydroxylase in Phenylketonuria
B. Dopamine Hydroxylase deficiency
C. Alkaline Phosphatase in Gushers disease
D. Purine metabolism in Lesch-Nyhan Syndrome
E. Ceruloplasmin in Wilson disease

78

Question: Which testing instrument is most applicable to test the intellectual functioning in a 4 year old child with hearing impairment?

A. Leiter-R International Performance Scale
B. Peabody Picture Vocabulary Test
C. Stanford–Binet Intelligence Test
D. Vineland Adaptive Behavior Scale
E. Wechsler Preschool and Primary Scale of Intelligence (WPPSI)

79

Question: A 10 year old mentally retarded male is on medication for ADHD. He has a long face, prominent large ears, hyper-flexible joints and macroorchidism. What is the likely diagnosis?

A. Fragile X Syndrome
B. Lesch-Nyhan Syndrome
C. Prader-Willi Syndrome
D. Turner's Syndrome
E. Williams Syndrome

80

Question: The Individuals with Disability Education Act (IDEA) was passed to accomplish which of the following?

A. Allow mentally retarded individuals to live in group homes and to receive education
B. Mandates free appropriate public education in least restrictive environments
C. Allow the mentally retarded to apply for social security
D. Allow the mentally retarded to decide which parent will obtain guardianship in case of parental divorce

81

Question: A newborn with an APGAR score of 9, undergoes standard testing for phenylketonuria (PKU) 12 hours after birth. The parents are relieved that the results are negative. The appropriate interpretation of these results is which of the following.

A. The negative results conclusively rule out the presence of PKU.
B. The test for PKU is valid only after the newborn ingests a diet containing phenylalanine.
C. The test for PKU has low sensitivity and standard protocol is to repeat it three times.
D. The PKU test is not done routinely in newborns in the US. It is done only when APGAR scores are below 7.

82

Questions & Answers

The End

83