Common Developmental Disabilities

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Learning Objectives

- Familiarity with the diversity of etiologies for common developmental disorders
- Develop a framework for primary care practitioner to use in initial assessment of children with atypical development
- Understand role of different specialists in helping to evaluate and care for children with common developmental disorders

Developmental-Neurodevelopmental Disorders

Intrinsic

Extrinsic

Multifactorial

Intrinsic Risks and Disorders

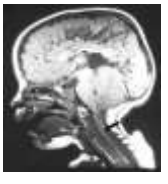
- Heritable disorders and errors of morphogenesis and metabolism
- Neuromotor disability
- Disorders of cognition and pervasive developmental disorders
- Higher prevalence conditions and learning disabilities
- Behavioral disorders

Errors of Morphogenesis

- 3% of all live-born infants have a major anomaly
- Additional anomalies are detected during postnatal life – about 6% in 2 year olds, 8% in 5 year olds, other 2% later
- Single minor anomalies are present in about 14% of newborns

Minor Anomalies

Major Anomalies



Definitions

Malformation

Deformation

Disruption

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Malformation

- Morphologic defect of an organ or region due to an intrinsically abnormal developmental process (e.g. hypoplasia, incomplete closure, incomplete separation)

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Causes of Malformation

• Multifactorial	20%
• Single-gene	7.5%
• Chromosomal	6%
• Infection	2-3%
• Maternal diabetes	1.5%
• Maternal medication	1-2%
• Unknown	>50%

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Congenital Malformation

- About 20-25% of perinatal deaths are due to lethal malformations
- Birthweight 500-1500 grams – 10%
- Birthweight >1500g – 50%

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Deformation

- Abnormal form or position of a body region caused by non-disruptive mechanical forces
- Examples:
  - Clubfoot
  - Congenital hip dislocation
  - Plagiocephaly

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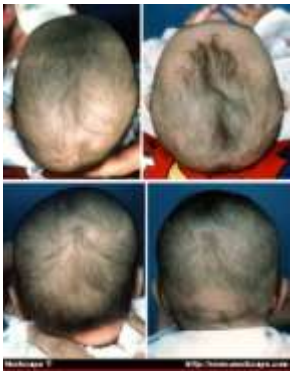

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Deformation Sequence

- Examples:
  - Intrauterine constraint
  - Robin sequence secondary to mandibular constraint

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Disruption

- Morphologic defect of an organ or region resulting from a breakdown of, or interference with an originally normal developmental process
- Example:
  - Amniotic Band Disruption

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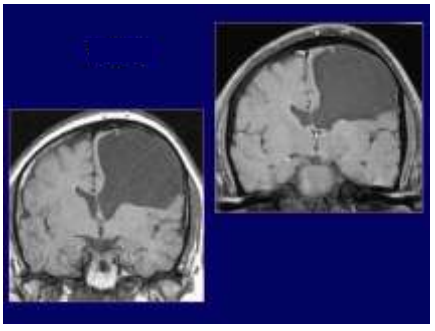
Porencephalic cyst



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Malformations of the neural tube (ex: Spina bifida)

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Neural Tube Defects

- Incidence
  - 2nd most prevalent congenital anomaly in U.S. (after congenital cardiac malformations)
  - 1-5/1000 live births
  - Girls affected more than boys
  - Ethnic and geographic factors remain important variables
    - Highest rates: Ireland, UK, Pakistan, India, Egypt
    - In U.S.: rates highest in East and South vs. West

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Neural Tube Defects

- Etiology
  - Multifactorial
    - Overall recurrence risk in U.S. with one affected offspring 1.5-3% (5.7-12% worldwide)
    - Syndromes (Roberts, trisomy 18)
    - Folate deficiency
    - Folate antagonists (phenobarb, trimethoprim)
    - Disruptions (amniotic bands, warfarin,)
    - Metabolic disorders (folate reductase' genes)
  - Genetics
    - High concordance in monozygotic twins
    - 7% of fetuses have aneuploidy (mostly trisomies)

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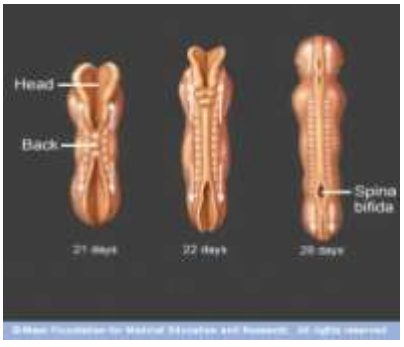

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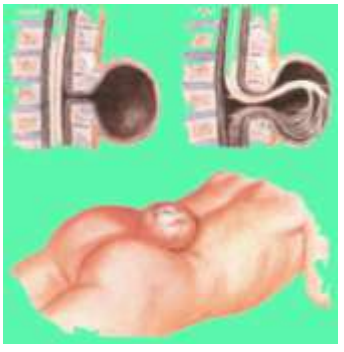

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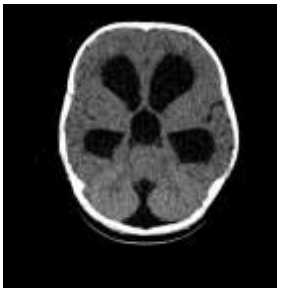
Hydrocephalus



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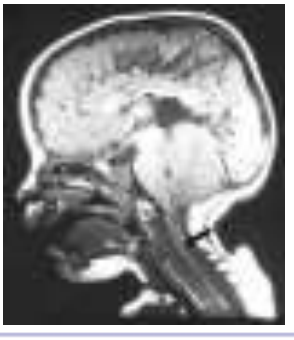
Chari II Malformations



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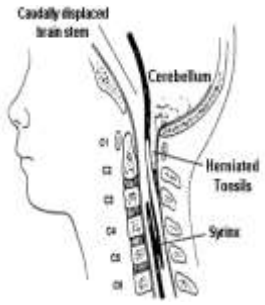
Chari II Malformations



- Herniation of medulla, tonsils, vermis
  - 4th ventricle at
  - foramen magnum
  - towering cerebellum
  - tectal beaking
  - myelomeningocele
  - aqueductal stenosis

  → hydrocephalus

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Chari II Malformations

- May present acutely: Central or obstructive apnea, aspiration, stridor, nystagmus and profound quadraparesis
- May progress to death regardless of type or acuity of treatment (usually <2 years old)
- Disorganized brainstem nuclei on autopsy
- May also present subtly: Hoarseness, dysphagia, pneumonia, increasing nystagmus, sensory changes in the upper extremities
- Shunt revision routine before decompression

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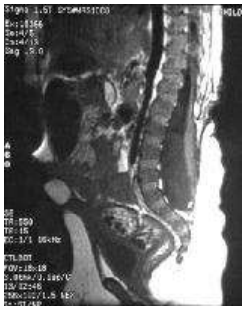
Tethered Cord



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Medical Care Priorities

- Early identification of AC-II symptomatology
- Latex allergy and atopy
- Developmental delay or mental retardation?
- Self-esteem, self confidence, infantalized behaviors
- Bowel and bladder independence
- Focused assessment of acute illness
- Transitional planning
- Adult independence

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Prognosis and Development

- 85% neonatal survival rate
- 72% of survivors are ambulating
- 87% have urinary continence
- T-12+ lesions associated with more severe CNS dysfunction
- 85% attending or have graduated high school and/or college (n=71)

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Inborn errors of metabolism and biochemical genetics

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Hurler Syndrome

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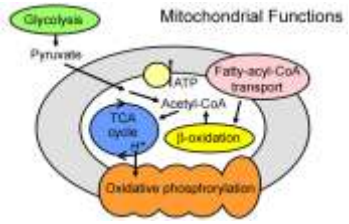

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Tetratogenic Disruptions

Teratogens are exogeneous agents that may cause developmental defects:

Drugs	warfarin; valproic acid; phenytoin; vitamin A; thalidomide; cytostatic drugs: cyclophosphamide, lithium carbonate
Chemicals	PCBs, methylmercury, alcohols)
Infections	rubella, cytomegalovirus, herpes, toxoplasma, syphilis
Ionizing radiation	RTG
Maternal factors	diabetes mellitus, hyperthermia, phenylketonuria, hyper-/hypo-thyreosis

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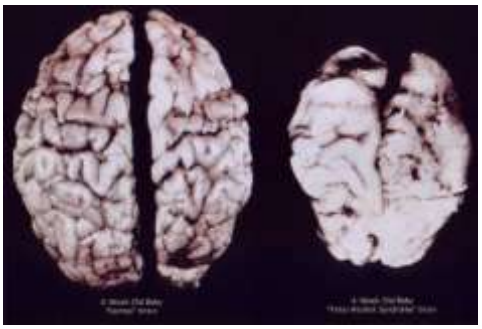
Fetal alcohol spectrum disorder (Ethanol embryopathy)



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Non-teratogenic Disruption

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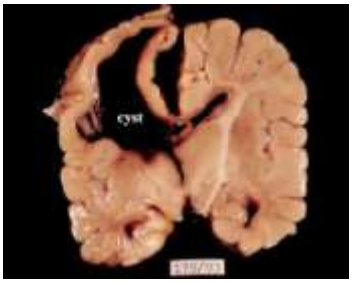

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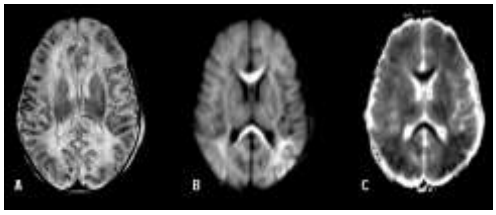
Severe hypoxia-ischemia



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Cerebral Palsy (CP)

- Term ~~c~~erebral palsy" often used as a diagnosis
- More precisely, however, it describes a motor manifestation of an underlying condition or event
- May not always present as spasticity
- Parents will benefit from having these distinctions made and clarified

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Cerebral Palsy—A Common Special Health Care Need

- 25,000 new diagnoses per year in the U.S.
- Milder forms, especially involving single or distal extremities may elude detection
- Incidence figures reflect varied thresholds for making the diagnosis

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Definition of Cerebral Palsy

- An impairment of movement and posture resulting primarily from either injury to or malformation of the cerebral cortex

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Patterns of Cerebral Palsy

. . . Impairment in movement and posture

- Increased tone (hypertonia, *usually* spasticity)
- Decreased tone (hypotonia, atonia)
- Altered movements (dyskinesia)
- Disregulated movement (ataxia)
- Combination/mixed pattern

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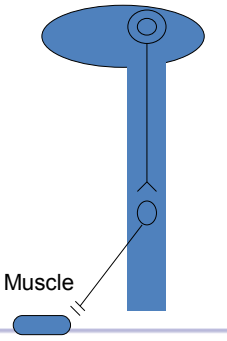
Basic Neural Pathway



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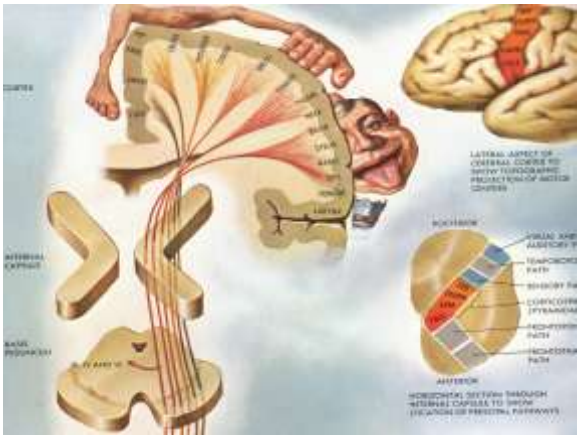
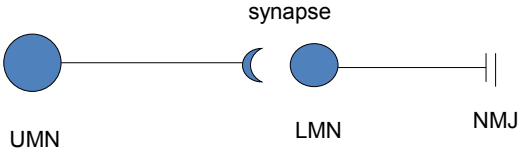


Upper Motor Neurons

Reside in the motor cortex



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Loss of UMN Function

- Without inhibitory action of UMN on LMN, tone becomes abnormal (typically increased/spastic), and uninhibited lower motor neuron function dominates reflex patterns
  - Hyperreflexia
  - Weakness
  - Increase tone

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Patterns of Cerebral Palsy

- RESULT:
  Ineffective voluntary movement, and a range of uncontrolled movements and reflexes which interfere with normal skills such as walking, sitting, and swallowing

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Epidemiology of CP

- Precise prevalence of CP is *uncertain*
- Overall incidence in U.S. estimated at 2/1000 live births
- Limited North American data on term infants since mid-1980's
- Rate of CP *is higher* for preterm infants and increases with decreasing birth weight and gestational age
- Worldwide incidence is elusive but may be higher than 2/1000

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Jarvis, S. et al Lancet, 2003

Epidemiology of CP 2004 data

- CP among 8-year-old children (N = 68,272) living in:
  - North central Alabama
  - Metropolitan Atlanta
  - South central Wisconsin
- Average prevalence of CP in 2004 across the three sites was 3.3 per 1,000

55

Disability and Health Journal, 2009 vol. 2(1): 45-48.

Epidemiology—Term Infants

- All congenital abnormalities
  - 19% of children with CP *vs.* 4.3% of controls
  - 14% of affected children had CNS abnormality
- CNS malformation/disruption…
- Intrauterine infection
- Multiple births
- Perinatal stroke
- Intracranial hemorrhage
  - 1/3 due to coagulopathy, thalamic hemorrage predominantly
- Acquired post-natal causes (10-18%)
- Kernicterus

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The case for hypoxic-ischemic encephalopathy as an etiology for Cerebral Palsy in children

- HIE accounts for 10-20 % of cases of CP
- Intrapartum events that *may* lead to asphyxia do not correlate strongly with CP
  - Abruption, prolapsed cord, placenta previa, nuchal cord
- Apgar score alone not strongly predictive
  - 7 year risk was 4.7% with 5 min Apgar of 0-3
  - . . . but 25x more predictive for scores of 7-10
  - 95% of infants with Apgar scores of 0-3 did not have CP
  - ? 10min+ Apgar scores may be more predictive

57

Nelson, K. Pediatrics, 1981; Paneth, N. J. Pediatr, 2001

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Other Considerations in Children with Cerebral Palsy

- Cognitive disorders (range of cognitive function)
- –Developmental delay" vs. –cognitive disability"
- Epilepsy—approx. 43% (range: 35-62%)
- Speech dyspraxia, speech/language disorders
- Strabismus, cortical vision abnormalities
- Gross motor dysfunction
- Fine motor dysfunction
- Self-care and vocational challenges

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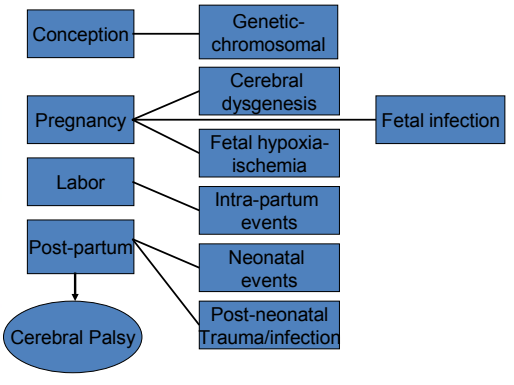

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Priorities in the Follow-up Care of Children with Cerebral Palsy

- Parent education
- Seating, positioning
- Ambulation
- Education
- Feeding/nutrition
- Social/emotional well-being

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Intrinsic Alterations of Gene Function Clinic Examples

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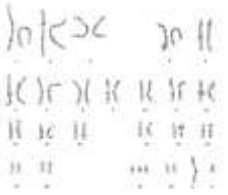
Down Syndrome



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Epidemiology of ASD

- All racial and socioeconomic groups
- Prevalence: 1 in 110
- M:F ranges from 3.2:1 to 7.6:1

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MMWR 2009;58 (SS10):1-20

Prevalence of ASD in WI—2006 Data

- Prevalence 7.6/1000 (1/132)
- About 5.5/1 Male : Female ratio
- Median age of diagnosis in Wisconsin is 53 months

67
MMWR 2009

3 Domains Affected

- Social Development
  - Less or no use of non-verbal behaviors
  - Less or no relating to peers
  - Less or no trying to share experiences, play with others
  - Less or no social or emotional reciprocity
- Communication Development
  - Less or no development of speech
  - Less or no trying to join conversation
  - Less or no social imitative or spontaneous make-believe play
  - Repetitive, stereotyped, or idiosyncratic language
- Behavioral Development
  - Very focused, restricted play
  - Repetitive non-functional behaviors
  - Stereotyped, repetitive motor mannerisms
  - Persistent preoccupation with parts of objects

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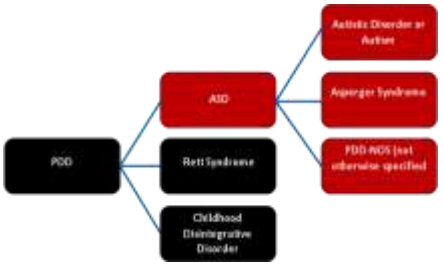
Pervasive Developmental Disorder (PDD)



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—Theory of Mind" (David Premack, 1998)

- Denotes the human ability to represent and make inferences about the mental state of one's self and others, encompassing desires, intentions, and beliefs

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Gassaniga et al., 1998

Theory of Mind

- The autistic child does not form a mental image of what can go on in other people's heads and this stems theoretically from a failure in thinking about his or her own mental states

71
Trevarthen et al., 1996

ASD Causal Genetic Variations

- 15q11-g13 region (also called the Prader-Willi/Angelman syndrome, or PW/AS region)
- Down syndrome, Smith-Magenis syndrome
- 22q13 deletion syndrome (SHANK3/PROSAP2)
- Rett syndrome MeCP2 gene
- Fragile X syndrome FMR1 gene
- Tuberous sclerosis TSC genes
- Smith-Lemli-Opitz syndrome DHCR7 gene
- Neuroligin genes (NLGN3 and NLGN4), ATR-X, and neurotrypsin

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ASD Susceptibility Loci

- More than 90 association studies have been published
- Chromosomes X, 2, 3, 6, 7, 10, 11, 12, 13, 16, 17, 19

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Rett Syndrome

- A progressive developmental disorder occuring in girls
- Multiphasic pattern of disability sharing features with autism
- Progressive debilitating

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Rett Syndrome—Necessary Criteria

- Normal prenatal and perinatal development, normal psychomotor development through first 5 months (5-18 mo)
- Normal head circumference; deceleration of head growth 5-48 mo.
- Loss of purposeful hand skills between 5-30 mo.
- Development of stereotypic hand movement (wringing, squeezing,clapping, tapping, mouthing, washing, rubbing)
- Development of severely impaired expressive and receptive language
- Evolution of severe psychomotor retardation
- Appearance of gait ataxia, truncal apraxia/ataxia between 1-4 years, social withdrawal
- Diagnoosis tentative until age 2-5 years

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Rett Syndrome

Rett syndrome *MeCP2* gene

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Aserperger Syndrome

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Pervasive Developmental Disorder-Not Otherwise Specified (PDD-NOS)

- DSM-IV-TR includes PDD-NOS in the category of ~~a~~utism"
- DSM-V may introduce changes

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Angelman Syndrome

15q11-g13 region

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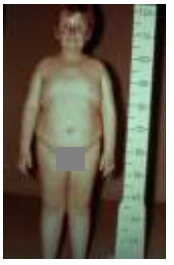
Prader-Willi Syndrome



15q11-g13 region

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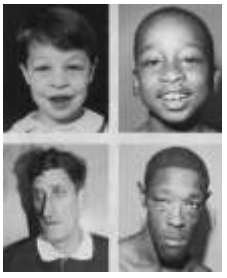
William's Syndrome



Fragile X Syndrome

FMR1 gene

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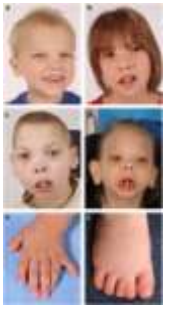
Smith-Lemli-Opitz Syndrome



DHCR7 gene

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Tuberous Sclerosis

TSC genes

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Cornelia deLange Syndrome

Gene linked to chromosome 3q26.3

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Diagnosis of Autism Spectrum Disorders

- No established medical ~~"tests"~~ in use to make the diagnosis of autism
  - ? eeg
  - ? Microarray
  - ? S-Amino acids, urine organic acids
  - ? High resolution chromosome analysis
  - ? *Fragile-X testing (PCR)*

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Intrinsic Alterations of Cortical Elaboration Clinical Examples

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Learning Disabilities

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Learning Disability

Specific problems in the acquisition and processing of information, old and new

- **Difficulties with attention**
  - ADD/ADHD
  - Anxiety
- **Difficulties with language processing**
  - Auditory processing disorders
  - Praxia (dyspraxia, apraxia)
  - Hearing loss and deafness
- **Difficulties with memory**
  - Visual memory difficulties
  - Auditory memory difficulties
- **Difficulties with visual processing**
  - Problems with vision
  - Problems in visual cortex
  - Motor problems impacting eye-hand coordination
- **Difficulties with visual/spatial organization**
  - Dyslexia
  - Dysgraphia
- **? Sensory processing difficulties**

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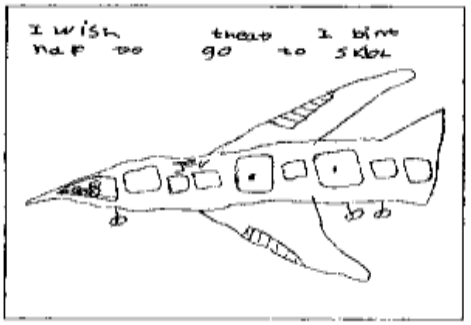
Dyslexia



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One out of 25 school-age children have dyslexia

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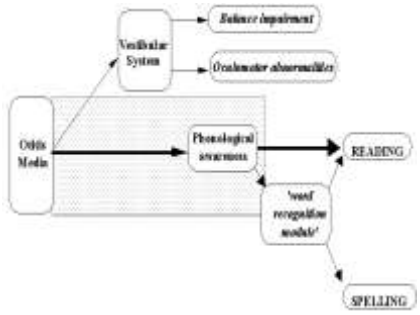

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ADD/ADHD

4% to 12% of school-aged children. About 3 times more boys than girls are diagnosed with ADHD.

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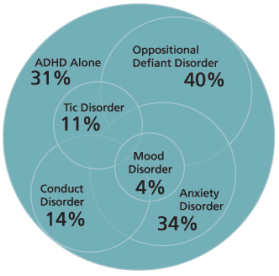
ADD/ADHD



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ADHD Measures

- ADHD-specific checklists
  - Connors Rating Scale
  - Swan, Nolan,Pelham-IV (SNAP)
  - Vanderbilt ADHD Teacher and Parent Rating Scales
  - ADHD Symptom Rating Scales
- Broad view of behavior tools
  - Behavior Assessment System for Children, 2nd ed. (BASC-2)
  - Achenbach Scales
- Research measures
  - Connors Continuous Performance Test
  - Test of Visual Attention (TOVA)
  - Gordon Diagnostic System

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Extrinsic Developmental-Neurodevelopmental Disorders

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Cognitive Disability

(Replaces the term ~~"Mental Retardation"~~)

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Definition of Intellectual Disability

Significant limitations both in intellectual functioning and in adaptive behavior, which covers many everyday social and practical skills. Originates before the age of 18.

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Intellectual Disability

- Intellectual functioning—(aka) intelligence
- Adaptive behavior (three skill types)
  - **Conceptual skills**—language and literacy; money, time, and number concepts; and self-direction
  - **Social skills**—interpersonal skills, social responsibility, self-esteem, gullibility, naïveté (i.e., wariness), social problem solving, and the ability to follow rules/obey laws and to avoid being victimized
  - **Practical skills**—activities of daily living (personal care), occupational skills, healthcare, travel/transportation, schedules/routines, safety, use of money, use of the telephone

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Intellectual Disability

- Other factors
  - **Community environment** typical of the individual's peers and culture
  - **Linguistic diversity and cultural differences** in the way people communicate, move, and behave
  - **Co-existent strengths**

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Intellectual Disability

Impairment in Intellectual Function

- Can be screened and suspected in infancy and childhood
- Definitively diagnosed after 5 years
- Diagnosis of made via formal cognitive testing and adaptive abilities assessment
- Often missed cause of anxiety, depression, school avoidance; other behavioral problems
- Irrefutably qualifies the child for an Individualized Education Program (IEP)

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Other Extrinsic Influences

- Bullying
- Conditions of low self-esteem
- Emotional trauma, abuse, neglect
- Others impacting mental health

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Ask a Question

Viewing online?

- Click on the chat icon above
- Question emailed to Training Team
- Questions answered by expert on topic
- Response within 2-3 weeks

Viewing at a live training?

- Organizer shares questions with Training Team

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Acknowledgements

- Wisconsin MCH LEND
- Department of Pediatrics, UW-Madison
- Wisconsin Medical Home Webcast Series
- Wisconsin CYSHCN Program
- Waisman Center

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