INTRODUCTION

1. Developmental disabilities or neurodevelopmental disabilities (includes intellectual disability)

2. Diverse group of chronic disorders (17% of those under 18 yrs in USA)

3. Begin at anytime during the development process (including conception, birth, and growth) to 22 years

4. Last throughout an individual’s lifetime
INTRODUCTION

1. Underlying basis for these disorders lies in fundamental deficits in the developing brain

2. Due to genetic, prenatal, perinatal, metabolic, and other factors

3. Affect 17% of individuals younger than 18 years of age in the United States

4. Prevention along with early recognition and intervention are critically important

5. Seek to mitigate the enormous personal and socioeconomic impact of these disorders
LIFETIME COST FOR THOSE IN US BORN IN 2000 WITH ID

1. Total cost: $50 billion
2. $11 billion for cerebral palsy
3. $2 billion for hearing loss
4. $2.5 billion for vision impairment

HISTORICAL PERSPECTIVE

1. Evolved based on a firm foundation of basic and clinical neurosciences
2. Father of developmental pediatrics, Dr. Arnold J Capute (1923-2003)
3. His fellows from the Kennedy Krieger Institute at John Hopkins University
4. Neurodevelopmental disabilities was recognized as a specialty by the American Board of Medical Subspecialties in 1999
5. Involves: Neurology with Clinical & Basic Neurosciences
6. Focuses on care of those with developmental disabilities across the lifespan
**Developmental Disabilities**

- Term emerged in US in 1960s
- Described functional impairment, not specific etiology
- Legal term as per public law 95-602 (1978)
- Does not include Mild MR
- Disability due to severe physical or mental impairment
Developmental Disabilities

- Identified under age 22 years
- Likely to be chronic indefinitely (lifelong)
- Considerable limits in 3 or more of areas of functioning
- Requires lifelong/extended care with multidisciplinary services
DEFINITIONS

• Framework of World Health Organization
• WHO Classification of Functioning, Disability, & Health
• 3 components to delineate disabilities
  --Body Function & Structure
  --Activity
  --Participation
• 3 interact w/ disorder + milieu + personal factors
  (WHO, International Classification, Geneva, 2001)
DEFINITION: Disability

- Global Term used to encompass problems with:
  - Body Functions
  - Body structures
  - Activity limitations
  - Participation restrictions

(WHO, International Classification, Geneva, 2001)
pediatric practice
Sports Medicine

Dilip R. Patel
Donald E. Greydanus
Robert J. Baker
Child with Disability
(http://idea.ed.gov)

- US 2004 Individuals w/ Disability Education Improvement Act (IDEA) that includes:
  - Intellectual disability (ID); Specific learning Disabilities
  - Hearing or visual impairments (incl deaf or blind)
  - Speech or language impairments
  - Serious emotional disturbance
  - Orthopedic impairments
  - Autism
  - Traumatic brain injury Other Health Impairments
Americans w/ Disability Act
(http://www.ada.gov)

- One w/ physical or mental impairment
- Limits one or more major life activities
- Incl caring for oneself, performing manual acts
- Seeing, hearing, eating, sleeping, walking
- Standing, lifting, bending, speaking, breathing
- Learning, reading, concentrating, thinking
- Communicating
- Working (6 months or more; not transient)
Intellectual Disability

- **Developmental Delay** (Correct sequence)
  --Sign. Delay of milestones/skills in 1 or more domains
- **Developmental Deviation**
  --Dev. Skills out of sequence
- **Developmental Dissociation**
  --Attainment of skills at sign. Rates between 2 or more
- **Developmental Regression**
Variety of terms used:
- Mental retardation
- Mental subnormality
- Mental disability
- Mental deficiency
- Feeble mindedness (late 19th century term)
Historical Perspective

• Long history of prejudice toward MR individuals

• *Mental retardation, epilepsy, and demon-possession:*
  – Confused in Western society for thousands of years
  – Identified as same process
  – All to be shunned and avoided

• Seen in Asian society: China as an example:
  – Zhou Dynasty (841 BC-221 BC) *(Su, 2005)*
  – Referred to MR child as “stupid, a child born stupid and fearful.”
  – 20th Century China: “handicapped” children allowed to die
In 16th to 19th Century Western Europe

- MR folks called *village idiots* and some lived as *court jesters*

- Called by Shakespeare as *Court Fools*: could speak their minds to the King, since considered to have no intelligence

- Lived as beggars or Church dependents; sent to asylums

- Most famous asylum at Bethlehem Hospital, called “Bedlam”

- Placing those with MR in institutions led to the concept of MR as a disease requiring medical attention
Other Laws

- **British Mental Deficiency Act** of 1913: placed in custodial care out of home
- 1927: US Supreme Court supported sterilization of MR females
- Nazi Germany enforced euthanasia for MR individuals (*Wolfenberger, ‘81*)
- 20th Century China: death to the handicapped child (*Su, 2005*)
- Change in the US: 1960: President Kennedy has sister with MR
1975: PL 94-142

• Education for all Handicapped Children Act
• Guarantee rights of institutionalized individual for quality services
• Mandated financial support be provided to the public education system
• Covered children with developmental disability (including MR)
• Must receive comprehensive education regardless of its severity
• Placed in the least restrictive environment possible
• Created public special education system in US
Individual with Disabilities Education Act (IDEA)

- Extended the range of services down to birth
- Formalized early developmental intervention systems nationwide
- Those with MR were removed from institutions
- Raised in their homes and learn vocation skills as adults
- As adults, stay in communities providing supervision
- Comprehensive support to help lead as normal a life as possible
Americans with Disabilities Act

- Made US law in 1990
- Expanded civil rights protection to all Americans with disabilities
  - Including developmental disabilities (Kastner, 1992)
- Public Law 94-142 was updated in 1997 to PL-105-17
- Emphasizing the right of parents to be equal partners with school officials in development of an individualized education plan (IEP) for their Child or Teen
DSM IV-TR, 2000

- IQ below 70
- Onset before age 18 years of age
- IQ tests used must be standardized and individually given
- 2 standard deviations below the mean
American Association on MR (AAMR)

- Redefined MR periodically since 1921 (Luckasson, ‘92)
- Combines below average IQ with:
  - Concurrent adaptive functioning impairment in **2 or more life skills:**
    - Health/safety
    - Functional academics
    - Work
    - Leisure
• Classifies MR according to patterns & intensity of needed support:
  – *Intermediate*
  – *Limited*
  – *Intensive*
  – *Pervasive*

• Adds environmental context—reflects WHO International views
MR Subtypes

- Subtypes of Mental Retardation (Curry, 2003; Carry, 2003; Patel/Greydanus, 2010)
  - **Mild**: 50/55 - 69 (about 3% of the population) ("Educable; Moron")
  - **Moderate**: 35/40 – 50/55 (about 0.4% of the population) ("Trainable; Imbeciles")
  - **Severe**: 20/25 – 35/40 (about 0.1% of the population have severe or profound MR)
    - Past term: Idiot
  - **Profound**: under 25/30
  - **Severity Unspecified**: Suspected, but not testable
Borderline Mental Retardation

• IQ above the mild MR range, but below the “normal” individual

• Affects about 7% of children and adolescents

• Often called “slow learners”

• Need special education services to maximize their potential as adults
Mild Mental Retardation (IQ: 50/55-69)

• 2.5%-3% of the population

• 85% (75%-90%) of individuals with MR have a mild type

• Sometimes classified as a “Familial” type of MR:
  – No demonstrable CNS pathology
  – Genetic or familial pattern is noted or inferred
  – In contrast to “Clinical” MR (moderate+): demonstrable CNS pathology
Mild MR

- Not distinguished from their peers until late childhood
- By late adolescence, have 6th grade academic skills
- As adults, they can possess acceptable social and vocational abilities:
  - Allow for self-support
  - With appropriate supervision and guidance
Associated Mental Disorders

• Most Common:
  – ADHD
  – Mood disorders
  – Pervasive developmental disorder
  – Stereotypic movement disorders
  – Mental disorder due to general medical condition (as head trauma – induced dementia)

(Greydanus & Bhave, Recent Adv Paediatr 2006; 17:174-92)
Aristotle to 21st Century

- Code of Hammuabi: 1750 BC
- Homer’s concepts: 1000 BC
- Jean Piaget: 20th Cent: Concrete & Formal Operational Thinking skills of older children & youth
- 21st Century MRI & Pet Scans: CNS matures in early to mid-twenties: Prefrontal Cortex
TEEN Brain: Early 21\textsuperscript{st} Cent Report

• Based on Neurobiology/N-Imaging
• MRI & Pet Scans of the CNS
• Most complex 1.36 KG universe mass
• Complex neuronal circuitry changes
• \updownarrow Grey matter and Pruning & Connects
• Suprachiasmatic nucleus of Ant Hypo
• Prefrontal cortex: Executive Function

US NIMH Longitudinal Brain Imaging Project (1989-)
Prefrontal cortex

- "area of sober second thought" / Decision making
- responsible for rational, executive brain functions / logic control
- skills as organizing thoughts, weighing consequences, assuming responsibility, interpreting emotions
- appreciation of fine music
- enhancement of motor skills
- **concrete operational thinking => abstract thinking** / Complex thinking
Neuroimaging Studies & CNS

- Anxiety circuitry preserved in species
- **Amygdala**: Fear conditioning and responses in all species
- **Hippocampus**: Contextual processing
- **Prefrontal cortex**: Fear modulation
- **PC**: Extinction of fear responses
21st Century: Brain Mapping to Identify Mental Illness

- **Cortex**: prefrontal and parietal
- **Brain stem**: reticular formation
- **Thalamus**
- **Basal ganglia**
- **Cingulate gyrus**
- **Limbic structures**: amygdala-hippocampus
Figure 1–3. A. Location of the central nervous system in the body. B. There are seven major divisions of the central nervous system: (1) cerebral hemispheres, (2) diencephalon, (3) midbrain, (4) pons, (5) cerebellum, (6) medulla, and (7) spinal cord. The midbrain, pons, and medulla comprise the brain stem.
Figure 1-9 (continued). B. Medial surface. The primary visual cortex is in the banks of the calcarine fissure.
Figure 1-16.  A. Drawing of the medial surface of the cerebral hemisphere.  B. Magnetic resonance imaging (MRI) scan of the midsagittal human central nervous system.  (B, Courtesy of Dr. Neal Rutledge, University of Texas at Austin.)
Pre-synaptic axonal terminal

Post-synaptic dendrite

Synaptic vesicle

Synaptic cleft

Release of neurotransmitter by exocytosis
BRAIN Neurochemistry: ERA of the Neurotransmitters in 20\textsuperscript{th} CENT

• Several Dozen Known NT in Humans

• May Indeed be Several \textit{Hundred} to Several \textit{Thousand} Unique Brain Chemicals

• “The Big Three:
  – Norepinephrine - 1946
  – Dopamine - Late 1950’s
  – Serotonin - 1940’s
Neurobiological Systems

- Noradrenergic
- Serotonergic
- Dopaminergic
- Cholimergic
- GABA (Gamma – Aminobutric Acid)
- Opioid
- Thyroid
- Glutamatergic
FIGURE 7  Serotonergic projection systems in the brain. The major serotonergic nuclei in the brain are the brain stem raphe nuclei (hatched). The nuclei are shown slightly enlarged and their diffuse projections (as described in the text) are markedly simplified.
MR: Differential DX/Co-morbidities

- Expressive Language Disorder (LD)
- Mixed Receptive-Expressive LD
- Phonological Disorder (Mutism)
- Borderline IQ
- Pervasive Developmental Disorder
- Dementia
- Schizophrenia
- Sensory Impairments

(Greydanus & Bhave, Recent Adv Paediatr 2006; 17:174-92)
MR Etiology

• MR represents a heterogenous group w/o a single cause

• Unknown in 50% (20-55%) w/ MR

• Cytogenetics and molecular testing advances will reduce unknown list
Etiology

• Among the identifiable causes:
  – Chromosome ABNL: Most Common
  – Aneuploidy: most common defect
  – Followed by abnormal X-linked genes
  – See 200+ genes causing X-linked MR
  – Poverty + malnutrition on mild MR
Main Etiologies for MR (Carey, 2003)

- MCA syndromes (multiple congenital anomalies)
- CNS malformation
- Metabolic disorder
- Acquired disorder
- Idiopathic (non-specific or pure) (20-55%)
Causes of MR
(Carey, 2003)

- Genetic: 50%
- Chromosomal: 30%
- Unknown: 25-38%
- Acquired: 15-20%
- CNS malformation: 10-15%
- MCA syndromes: 4-5%
- Metabolic: 3-5%
Causes in Children with Severe MR

- Chromosomal Disease (as Downs Syndrome): 22%
- Genetic Syndrome (as Fragile X Syndrome): 21%
- Developmental Brain Abnormality (as Lissence Phaly): 9%
- Inborn Error Metabolism (Mucopolysaccharidoses): 8%
- Neurodegenerative Disease
Comments on Etiology

- Specific cause is typically found in the severe MR types: 75%
- Identified cause is usually biological, psychosocial, or both
- Etiology is the same among SES: except in poverty-related:
  - Increase in lead poisoning and premature births
- Mild MR is often a combination of environmental and heredity factors
• Teen is at the lower end of the IQ spectrum because of genetic factors

• Mild MR is 4Xs more likely in offspring of females w/o HS grad

• Reflects on the influence of: (Shapiro, 2004)
  – Inherited cognitive dysfunction
  – Poverty
  – Larger families
  – Undernutrition
The More Severe the MR, The Greater Risk There is for

- Seizure activity
- Sensory handicaps (ie, visual or auditory)
- Motor disorders (as spasticity)
- Other neurological disorders
- Cardiovascular conditions
Evaluation

• General Principles
  – MR is OFT DX by Teen years
  – Mild MR DX in early Teen Years
  – Some may enter adolescence without an established diagnosis
  – No etiology attempt in many
• Diagnosis more difficult in Teens:
  – Effects of age on physical features
  – Changes in the family structure

• Advances in genetic testing over past decade:
  – Some MR teens: NO careful analysis
  – Not all children/teens have equal access to a full evaluation with modern technology
• Children with MR evaluated for:
  – Academic dysfunction
  – Behavioral problems

• Anxiety

• Reduced attention span
  *(Fragile X or Fetal Alcohol Syndromes)*

• Increased conduct problems
Fig. 1. The hyperactive adolescent at home.
ADHD/ADD

• Neurobehavioral disorders
• Abnormalities in neurotransmitter systems
  – Noradrenergic
  – Serotonergic
  – Dopaminergic
• Characterized by:
  – Attention dysfunction
  – Impulsivity
  – Hyperactivity
Comorbidities Commonly Found in Adolescents With ADHD

- Disruptive behavior disorders
- Mood disorders (unipolar and bipolar)
- Anxiety disorders
- Tourette syndrome and chronic tic (less commonly)
- Learning disabilities and executive function deficits

Medical Disorders

- Hyperthyroidism
- Subclinical epilepsy
- Fontal lobe tumor or abscess
- Fetal alcohol syndrome
- Klinefelter syndrome
- Angelman syndrome
- Williams syndrome
- Velocardiofacial syndrome
- Sotos syndrome
- Meds effects: phenobarbital, amphetamines
ADHD in older Teens & Adults

Inattention tends to persist into adolescence and adulthood
- Difficulty sustaining attention\(^1\)
- Paralyzing procrastination\(^1,2\)
- Disorganization\(^2\)

Impulsivity in adulthood may have serious consequences
- Impatience/losing temper\(^1\)
- Quitting/losing jobs\(^1,2\)
- Ending relationships\(^1,2\)

Hyperactivity often changes to inner restlessness\(^3\)
- Self-selection of an active job\(^1,2\)
- Fidgeting\(^1\)
- Excessive talking\(^1\)

FIGURE 2. Facial shape in Down syndrome, illustrated from 10 months to adolescence in a single person.
DOWN SYNDROME

• Maladaptive, Externalizing BEH
• Depression
• Seizure Disorder (10%)
• Alzheimer’s with aging
Angelman Syndrome

- Genetic disorder: loss of normal maternal contribution to a region of chromosome 15 (segment deletion) (absence of UBE3A gene)
- Prader-Will: similar loss of paternally-inherited genes
- Named after British Pediatrician (Dr. Harry Angelman) in 1965
- Intellectual/Dev delay, seizures, jerky motions, happy demeanor

- Sleep problems (May improve during adulthood)
  - Short attention span and easily distraction
  - Mild to Moderate developmental delay
  - Seizures and anti-epileptic meds
Angelman Syndrome
Characteristics Change From Childhood to Teen Years: Angelman Syndrome

• Severe developmental delay and seizures as a child

• Additional features as a teen:
  – Ataxic gait
  – Uncontrolled laughter
  – Frequent smiling
FIGURE 10. Facial shape in Noonan syndrome, illustrated in infancy and adolescence in a single person.
Beckwith Wiederman Syndrome

- Rare overgrowth disorder with various features
- Names in 1964 and 1969 (Germany & USA)
- Linked to chromosome 11 (arm 11p)
- Obstructive apnea due to large protruding tongue (macroglossia)
- Fatigue due to low blood sugar
- Sleep dysfunction due to general depression
- Non-specific Sleep dysfunction
FIGURE 4. Facial shape in Beckwith-Wiedemann syndrome, illustrated from birth to adolescence in a single person.
Rubinstein-Taybi Syndrome

- Rare autosomal dominant disorder; 1963
- Haploinsufficiency of CREBBP gene product
- Distinctive facies, short stature, mental retardation
- Broad, angulated thumbs & great toes; GERD
- Cardiac and renal defects; tumors; Resp IFs
- **Sleep apnea** often found; Obesity is common
- Non-specific Sleep dysfunction also seen

(Roelfsema, 2007; Wiley, 2003)
FIGURE 7. Facial shape in Rubinstein-Taybi syndrome, illustrated from birth to adolescence.
Health Problems with Mild MR/DD

- Similar to youth with a normal IQ
- Important concept
- W/ proper education & guidance
- Can lead a productive, happy life
Primary Care Clinicians & MR Prevention (Shapiro, 2002)

- Counseling youth about avoiding prenatal drug/alcohol abuse
- STDs (including HIV) prevention--including during pregnancy (Anni, 2002)
- Prevention of unwanted adolescent preg
- Provision of maternal folic acid supplementation for pregnancy
Pediatricians Should Actively Deal With DD Teens

- Inquire about:
  - Potential social isolation
  - Sexual behavior
  - Academic difficulties

- Be part of a multidisciplinary team helping adolescents
Need Help With Their Limitations in Dealing With Various Stressors

• Social isolation
• Peer rejection
• Constant failures
• Potential restrictions imposed by parents
Importance of Social Skills
(Bielecki, 2004)

• Dysfunction in cerebellar neural pathways (Cornish, 2005)
• SS deficits and excesses are important part of Teen with MR’s life
• See link between social skills and maladaptive behavior
• Training can reduce mental health complications
Adolescent Sexuality
(Greydanus, 2008)

• Same sexual desires and fantasies that all youth have

• Leads to increased risks for:
  – Sexual behavior
  – Sexual assault
  – Sexually transmitted diseases
  – Pregnancy
PLATO
(427 BC -347 BC) (The Republic)

• The son feels equal to his father
• He has no respect for his parents anymore
• All he wants to be is free
• Students insult their teachers
• And on top of this situation, in the name of liberty and equality…..
• SEX IS EVERYWHERE!
Robert Lewis Stevenson
Classic Teenager

• Long hair, different dress from parents
• Rejected parent’s religion
• Visited brothels
• “What a curse I am to my parents; My father said: “You have rendered my whole life a failure!’ My mother said: ‘You are the heaviest affliction to befall me.’””
Adolescent Sexuality
(Greydanus, 2002; 2008)

• Complicated by often limited sexually education provided to these youth

• Need education: inappropriate sexual touching and abuse

• Address GYN needs of females

(Greydanus, Peds Clin No Amer, ’02; ’08; Dis A Mon ’09: 55: 1-60)
Factors Complicating GYN Care

- Communication difficulties in DD PTs
- Cognitive limits that may be found
- Increased neurological problems: seizures
- Multiple joint contractures: spasticity
- Kyphoscoliosis; autonomic dysreflexia
- Nutritional issues: feeding tubes, GE reflux
- Clinician refusal to consider GYN issues
- Limited clinician’s training

(Greydanus & Omar Menstrual Dis in Adol Females. Dis A Mon ‘09: 55: 1-60)
Factors Complicating GYN Care

- Clandestine clues in those with comm diffic.
- UTI: crying w/ urination + foul smelling urine
- UTI: Fever without obvious origin
- Excessive vulvar irritation: masturbation
- Vaginal discharge post antibiotics: yeast IF
- T vaginalis in urine/Pap smear: Sexual abuse
- Purulent vaginal discharge: Foreign body
- Non-specific vulvovaginitis in female children
- Specific VV: Streptococcus, Shigella, others

(Greydanus, Peds Clin No Amer 2002; 13:223; Dec, 2008)
Prognosis

• General concepts
  – Medical stabilization OFT by TEENS
  – Behavioral issues may develop as a complication of adolescent DEV.
  – Depends on level of IQ & Support
    • Education
    • Management Opportunities
Transition to Adult Life and Care

- Transfer to adult health care as per specific country health care system
- Need for uninterrupted prim/spec care
- Acquire knowledge for adult living
- Need for accommodations for adult life
- Sexuality needs (gynecologic for females)
- Mental health care needs

Major Barriers to Successful Transition

• **Low expectations & social isolation**
• Adult care provider w/o DD care training
• US: Teen resistance to enter adult care
• US: Family resistance to transition
• US: Lack of reimbursement for care
• Lack of coordinated multidisciplinary care
• Sexuality and gynecologic needs
• Mental health care needs

Health Care Screening in Aging Adults with DD

- **Cardiovascular and Cancer Screening**
- Cancer: breast, cervix, colon, prostate, skin
- Chronic obstructive pulmonary Disease (COPD)
- Dimentia and Alzheimer Disease
- Diabetes mellitus
- Lipid Screening
- Hearing and vision (glaucoma) screening
- **Immunization Status**
- Malnutrition
- Abuse: Sexual, Physical, Neglect
Health Care Screening in Aging Adults with DD

- **Mental Disorders: Depression, Anxiety**
- Obesity
- Osteoarthritis
- Osteoporosis
- Periodontal Disease
- Podiatric Problems
- Spinal Disorders
- Sexual Dysfunction
- Sexually Transmitted Diseases (Viagra syn!)
- Peri-Post Menopausal Issues
Use of Psychopharmology

• Higher prevalence of mental health disorders

• Co-morbid mental health issues may hinder success in:
  – Social skills attainment
  – Housing and employment procurement

• Limited research on Psychopharm effect
Current research indicates that 10 to 20 percent of children and adolescents suffer from mental health problems at some point before reaching adulthood. Complex diagnostic criteria and medication regimes can make these patients a challenge to manage. The chronic shortage of child and adolescent psychiatrists has led to more and more cases being cared for by pediatricians and primary care practitioners. Patients may be on multiple medications for an extended period, and reaching a decision to modify or cease treatment is often complicated.

Pediatric and Adolescent Psychopharmacology: A Practical Manual for Pediatricians, is written for all non-psychiatrists who find themselves caring for children and teenagers with mental health disorders. It provides practical, accessible advice on the complex psychopharmacologic management options available for many mental health disorders of children and adolescents. This text will be an invaluable resource for all clinicians who encounter these vulnerable patients.

Other titles of interest
Adolescent Substance Abuse
Udle and Rowe
(ISBN 9780521823586)
A Clinician’s Handbook of Child and Adolescent Psychiatry
Gilberg, Harrington and Steinhausen
(ISBN 978052119367)

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• **Michelangelo** (1474-1563 AD) (David, Adam)
  *Ancora imparo* (I am still learning)

• **Seneca** (63 AD) (124 letters at end of his life)
  *(Epistulae morales ad Lucilion)*
  *H omines, dum docent, discunt*
  *MEN LEARN WHAT THEY TEACH*

• **Aristotle** *(Nicomachean Ethics)*
  *Happiness is once the best, the noblest*
  *And the Most Pleasant of Things!*

• **Plato** *(The Republic)*: *Those having torches will pass them on to others.*
Help all Children Reach Productive Adulthood