

# A community-based developmental pediatrician's perspective on evidence-based priorities in ASD- management

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# Overview

- Provide a historical perspective of autism and diagnosis of ASD
- The “system” for the diagnosis and care of children with ASD.
- General pediatrician’s role/ shortcomings with the system.
- Developmental pediatrician’s role and approach to diagnosis and care of children with ASD
- My perspective on research priorities

# Autism: What is known?

- We can reliably identify children at high risk long before age 3, and long before the development of functional language.
- Boy : Girl ratio ~ 4:1
- No definitive cause, therefore no effective means of prevention, and no specific or targeted treatment for “cure”.
- Early identification/intervention IS effective and improves prognosis by reducing disability.

*[www.nichd.nih.gov](http://www.nichd.nih.gov)*

# History of autism

- 1943: Kanner –  
*Autistic Disturbance of Affective Contact*
- 1940-1950s: Childhood Schizophrenia brought on by aloof (“refrigerator moms”) parenting (Kanner & Bettelheim).
- *refuted* by all attachment studies

# **Autism: Behavioral Perspective**

- **1960s: Autism as a Behavioral disorder.**
- **Treatment focus on behavioral conditioning strategies to reduce maladaptive behavior.**
- **Less emphasis was placed on developing social and communication skills.**

# Autism: Cognitive Perspective

- **1970s : Autism as a cognitive disorder with defects in:**
- **Theory of Mind (Defect in Empathy; “Mind Blindness”).**
- **Executive Dysfunction (Defect in planning action/defect in integrating sensory input)**
- **Weak Central Coherence (Defect in Integration; focus on parts rather than the whole)**

# Autism: Current Thinking

- Autism is a complex and highly variable Neurodevelopmental disorder.
- **Has Distinctive Behavioral and Cognitive Abnormalities and a strong genetic influence.**
- **This “Biologic Basis of autism” helps explain**
  - Similarities in presentation and cross-cultural prevalence; common changes in brain structure and function; and common associated systemic involvements (GI, Allergy, sleep disturbances).



# Evidence for Autism as a Genetic Disorder

- Twin studies
- MZ (Identical twins): 70-90% have concordance for autism spectrum.
- DZ (fraternal) twins have same risk as sibling recurrence (4-8%)
  - 20-100 fold increase over general population risk

# Evidence as a Genetic Disorder: ASD in congenital syndromes

- 3-9% of ASD-individuals have an associated congenital syndrome w/ chromosomal abnormalities. Some experts believe that studying these genetic abnormalities may shed light on ASD-pathogenesis.
  - 15q duplication (1-3%)
  - Fragile X (1-3%)
  - Tuberous Sclerosis (0.5-3%)
  - Rett's syndrome

# Many rare genetic diseases are associated with Autism

- Fragile X
- Tuberous sclerosis complex
- Neurofibromatosis I
- Hypomelanosis of Ito
- Down syndrome
- Prader-Willi/Angelman
- Untreated PKU
- Disorders of purine metabolism
- Williams syndrome
- Sotos syndrome
- Smith-Lemli-Opitz syndrome
- Noonan syndrome
- Joubert syndrome
- Congenital myotonic dystrophy
- Duchenne muscular dystrophy
- Leber's congenital optic atrophy (mitochondrial)

# The Autism Spectrum

- DSM-IV described the Autistic Spectrum as a group of related disorders ranging from mild to severe disability which include the following:
  - **ASPERGER SYNDROME**
  - **ATYPICAL AUTISM/ PERVASIVE DEVELOPMENTAL DISORDER (PDD-NOS)**
  - **AUTISTIC DISORDER (Classic Autism)**
  - **CHILDHOOD DISINTEGRATIVE DISORDER**
  - **RETT SYNDROME**

# Shift in DSM-IV versus DSM-V

- In 2013, the DSM-V has replaced DSM-IV.
- The DSM-V merges the language and social domains, and streamlines the diagnosis.
- The terms PDD-NOS and Asperger's syndrome have been removed from the ASD diagnosis.
- ASD are defined by characteristic abnormalities in social communication and interactions and presence of restrictive repetitive behaviors **(sensory included)**

# The “System” in “Early Identification and Intervention”.

- **FACT:** Early identification and intervention alters developmental trajectories, and decrease morbidity\* in ASD.
- General Pediatricians (GPs) are well positioned to assess for developmental referral.
  - Trusted source for information
  - Frequent access (6-10)
  - GPs increasingly aware of ASD-risk factors and features

\*Warren et al, 2011 Pediatrics; Brian et al, 2015 Current opinion in neurology

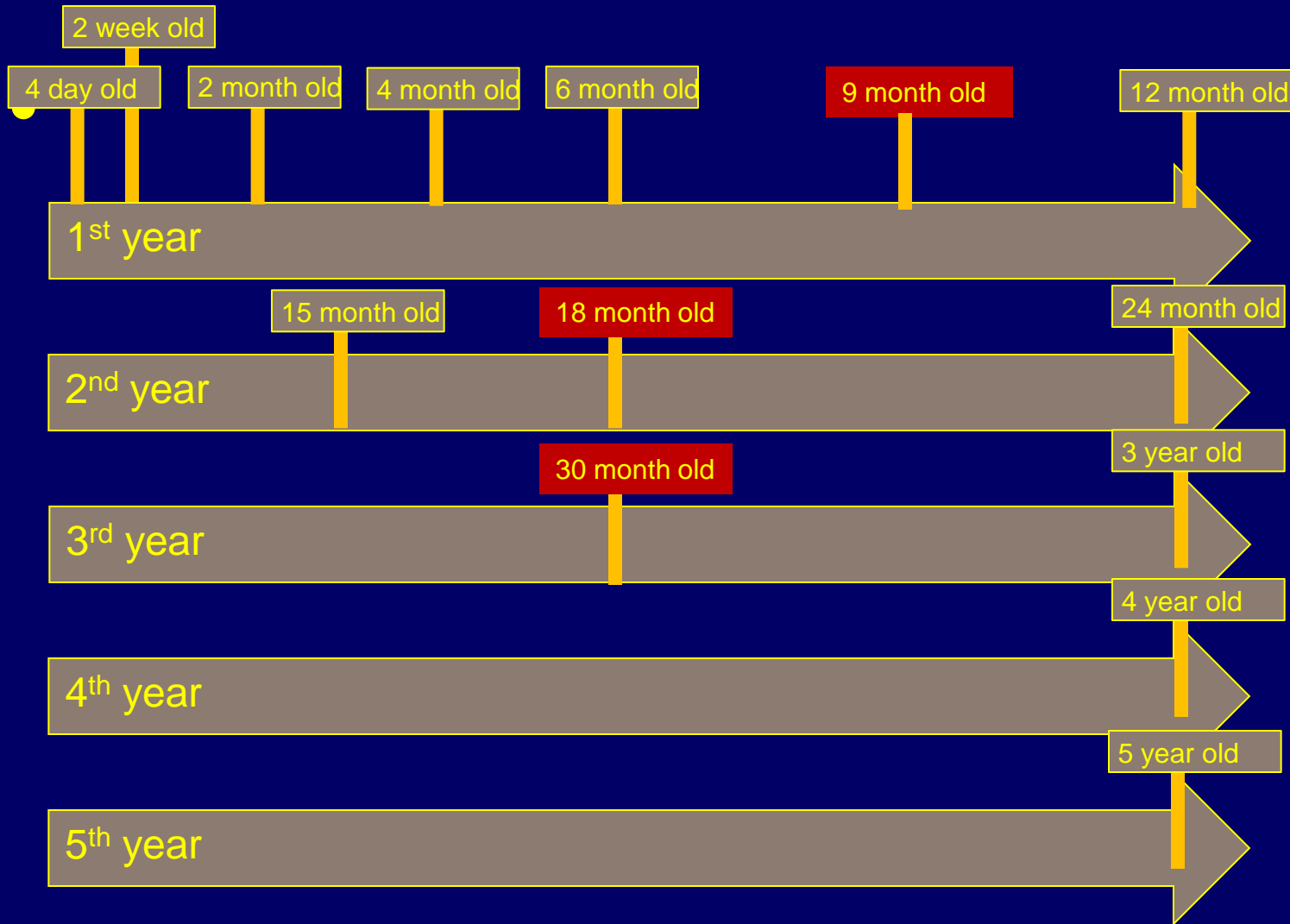
# The General Pediatrician (GP)

- Provides comprehensive medical care for infants, children and adolescents
  - Safety and injury prevention (car seats, helmets, vaccinations)
  - Respond to acute conditions (colds, sprains, ear infections)
  - Manage chronic conditions (asthma, DM, ADHD)
  - Anticipate developmental issues (breast feeding, safety, starting solids)
  - Screen for potential problems (Review of Systems, growth charts, developmental delays).



# Well Care Visit Schedule

Skewed towards infant care and vaccines





# Pediatric Guidelines for Developmental Screening

- AAP recommends developmental surveillance at all well-care visits
- GPs are to address concerns promptly, or refer for more in-depth evaluation
- Developmental screening tests at the 9-, 18-, and 24/30-month visits (not paid by insurance)
- M-CHAT at 18 and 24 month visits



# Challenges in Well Care and Developmental Screening

- Visits to GPs “sick visit” Vs. “well care”.
  - GP time is limited – Healthcare Systems allocate 15-20 minutes for MD allotted for a “check up”.
  - Screening for Developmental Issues may not reach high priority.
  - Screening for Developmental Issues is time-consuming
  - GP-knowledge base for conducting developmental screens may be limiting.
  - Reimbursement is limited.



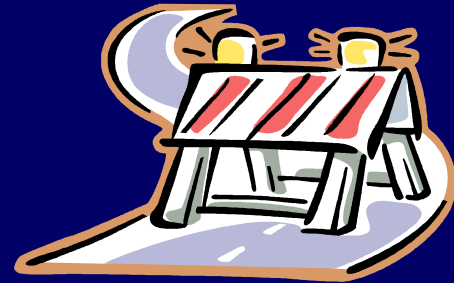
# GP Screening: Developmental Assessment needs to be a Priority



- Over 40% of parents of children aged 4m-5y expressed at least 1 concern about their child's physical, behavioral or social development according to a 2007 survey <sup>1</sup>
- Nearly 14% of children 24 m/o have developmental delays that are likely to make them eligible for early intervention services by IDEA <sup>3</sup>
- Over 40% of parents in 2000 reported that their 35 month old had never received a “developmental assessment” <sup>2</sup>

# Barriers to Pediatrician Screening

- Limited time
- Limited training
- Limited reimbursement
- Need proper measures (fast, easy, cheap, good metrics)
- Patients need access to care in the first place



# “Clinical Judgment”

- Historically pediatricians have relied on clinical judgment to evaluate developmental and behavioral concerns



Silverstein, *Pediatrics* (2005)

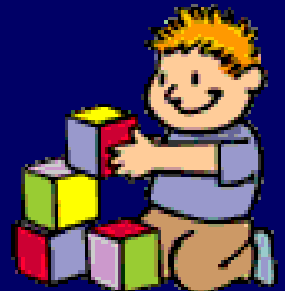
# The GP-Clinical Judgment often doesn't elicit threshold concern

- Lack of standardization (subjective)
- Limit of child cooperation (“white coat syndrome”)
- Clinical judgment identifies <30% of children with developmental disabilities\*.
- And easy, validated, time-efficient tests for “age-specific developmental delay” are generally lacking\*.

\*Silverstein, *Pediatrics* (2005); Glascoe, *Pediatrics* (1995)

# Developmental Screening Tools

- Denver II (1990)
  - Assesses 4 domains (personal-social, fine motor, gross motor, language)
  - 2wks – 6 years old
  - **15-30** minute test
  - **However, accuracy limited**
    - good sensitivity (83%) but low specificity (43%), miss mild delay and language difficulties



# Developmental Screening Tools

- Ages and Stages: **Milestone questionnaire**
  - (4-60m/o, 70-90% sensitive, 76-91% specific) milestone oriented

## **GROSS MOTOR** (continued)

4. Does your child jump with both feet leaving the floor at the same time?



5. Does your child walk up stairs, using only one foot on each stair? (The left foot is on one step, and the right foot is on the next.) He may hold onto the railing or wall. (You can look for this at a store, on a playground, or at home.)



6. Does your child stand on one foot for about 1 second without holding onto anything?





# Developmental Screening Tools

- PEDS: (Parents' Evaluation of Developmental Status).
  - Concern oriented questionnaire
  - birth-8y/o, 74-79% sensitive, 70-80% specific

Do you have any concerns about how your child uses his or her arms and legs?

Circle one: No Yes A little COMMENTS:

*He's very coordinated and very fast!*

Do you have any concerns about how your child behaves?

Circle one: No Yes A little COMMENTS:

# Parent Concern: Screening Tool

- Parent concern will identify 80% of children who fail screening tests\*
  - More predictive for speech and language, fine motor skills, general functioning.
  - Less with self-help, gross motor, behavior
  - Much better with current skills than historical recall
- Screening questionnaires designed to capture parent concern



\*Silverstein, *Pediatrics* (2005). Glascoe *Pediatrics* (1998).

# Parent Concern has to be translated into language a doctor can understand

- Important to ask questions properly
  - “..any concerns about the way your child is learning, behaving, developing...”
  - “does your child use any of the following words?”
- Parental report is often more sensitive than professional assessment
  - Emerging, inconsistent skills are more often demonstrated at home than over a 20 minute visit in a doctor’s office.

# Parent Concern: Caveats

- Varied experience with young children
- “Anxious parenting”
- Lack of typical/atypical comparisons
- Parental concern : denial vs anxiety vs stigma
- Cultural factors (expectations, bringing up concerns)
- “Guilt” (Varied amount of time parents are able to spend with their kids).



# Autism

## Consequences of Missed or Misinterpreted Screening

- Average age at time of diagnosis can be as late as 3 ½-4 years (dependent upon community and practitioner awareness).
- Minority (underserved) communities in the USA are often diagnosed much later.
- Average age of initial parental concern is 14-18 months.

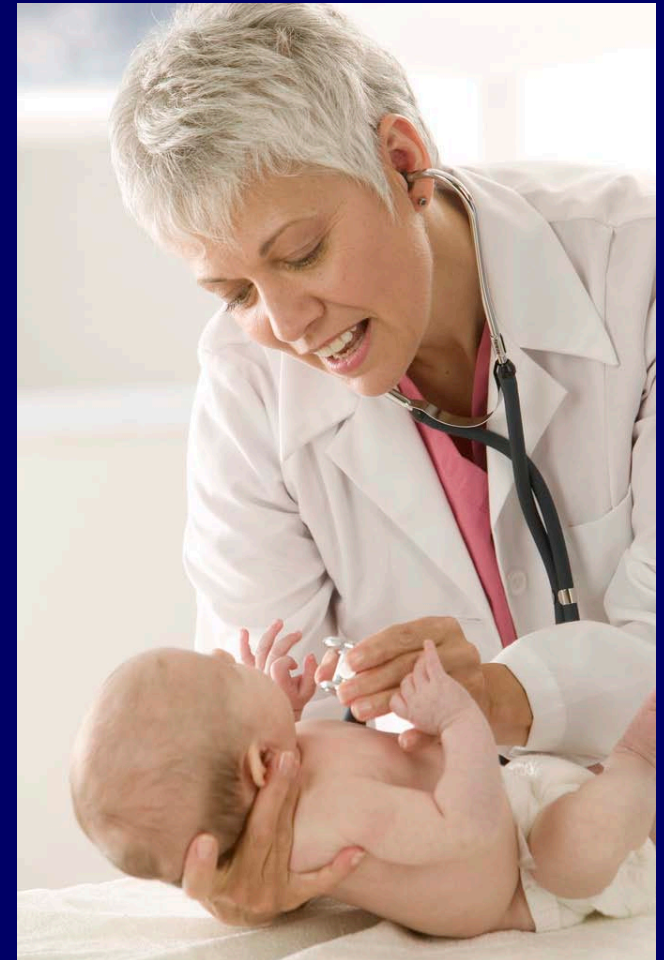
(Chawarska K. et al J Aut Dev Dis. 2007; 37:62-72)

# The Bottom Line for GP-assessment!

- All “measures” of development are tools for risk assessment. No test is perfect!
- GP-job is to do them, make sense of them and decide:
  - Refer to Developmental Pediatrician (DP)
  - Monitor (wait and see)
  - Counsel parents on interventions
  - Reassure that their child is developing “normally” (typically).

# What is Developmental-Behavioral Pediatrics (DP)

- Subspecialty formally recognized and boarded by the ABP in 2002
- 3-year residency in Pediatrics
- 3-year fellowship-focus
  - Child Psychology/Psychiatry
  - Neurology
  - Genetics



# DP-foundations

- Rooted in the study of early child development
  - Darwin – “An evolutionary Biological Sketch of an Infant”
  - Freud – psychosexual, subconscious, childhood experiential development.
  - Gesell – maturational theory, biologically based
  - Piaget – children think differently, clinical method, developmental stages
  - Vygotsky – sociocultural perspective
  - Bowlby – attachment theory



# Developmental Behavioral Pediatrician Role

- Evaluation / “Demystification”
  - Developmental / Psychological Testing
  - Medical / Genetic Testing
- Interdisciplinary Care-”Captain” of the team
- Advocacy and Family support
- Management and ongoing Follow-Up
  - Referrals
  - Therapy
  - Medications

# Developmental Behavioral Pediatrician Role

- Not general pediatric visit (don't give vaccines or treat ear infection)
- Subspecialty neurodevelopmental evaluation and ongoing management, referred to by general pediatricians, therapists, teachers, etc.
- Office is “non-threatening”; homey with toys and games – allow children to feel comfortable and demonstrate their true abilities

# Developmental Pediatrics Evaluation Process

- Evaluations vary by age and concerns
- **Assessment usually 2-4 visits, each visit usually 1.5 – 2 hours**
- Meet with parents **for intake and feedback**
- **Spend time doing evaluations with children** (school, play, home)
- **Speak with therapists, teachers, other team members, review of previous records**
- **To obtain overall “big picture” of child, strengths and challenges**

# Comprehensive Historical Diagnostic Evaluation

- Thorough Pregnancy, Birth and Developmental History
  - age of mother, perinatal “injury”, assistive reproduction, prematurity, post natal temperament, “spitter”, sleep, nutrition
  - emphasis on motor development and visual attention. Focus on level attainment and/or regression,
- Family History-“genetic salt shaker”
- Medical History (vaccinations, GI-complaints, Allergies, recurrent ear infections, medications and environmental exposures).

# Comprehensive Physical Diagnostic Evaluation

- Physical and Neurological Exam
  - Head Circumference, growth curves, height/weight, skin lesions (NF or TS), dysmorphic features, Muscle Tone, Posture, Gait and Reflexes.
- Direct Observational Evaluation (office and school)
  - Parent-Child, Child-Adult, and Peer-Peer interactions

# Developmental Testing

- Assessments look at:
  - Development (GM, FM, EL, RL, VR)
  - Social Communication
  - Achievement and Learning
  - Attention and Executive Function
- Interpretation – quantitative and qualitative

# Other Testing

- Achievement – WIAT-III, WRAT-4
- Attention – Gordon, TOVA
- Language – Expressive/Receptive 1-Word Vocabulary Tests
- Visual Motor – Beery VMI
- Sensory – Sensory Motor Profile

# Autistic Spectrum Testing

- Autism Diagnostic Observation Schedule (ADOS)
  - 1 hour observational test, 4 modules by age / language skill
  - Give opportunity to show social communication skills
- Autism Diagnostic Interview (ADI-R)
  - 3 hour interview of parents, 93 items
  - For older child, ask current traits & traits at 4-5y/o



# DP-Toolbox for Developmental and Screening Evaluation

- M-CHAT (5 minute)
- M-CHAT Interview (10-15 minute)
- Comorbidity evaluations (ADHD: Vanderbilt, Connor's, SNAP); (Adaptive / Personal Social Skills: Vineland Adaptive Behavior Scales); (General Behavior Screening: Child Behavior Checklist (CBCL), Pediatric Symptom Checklist).
- Developmental Schedules (Mullen 1995; Bailey Infant Development 2006; Gesell revised 1980; McCarthy Scales 1972; Battelle Developmental Inventory 2004)

Kleinman, J of Autism Dev Disord (2008)

# Diagnostic Evaluation Medical testing may include.....

- Auditory & functional vision evaluation
- EEG- Rule out seizures, 24 hour if possible
- Metabolic Screening/Mitochondrial disorder:  
Blood, Urine, carnitine, coq 10, “mito”-cofactors
- Genetic testing: microarray, Fragile X, Retts
- Neuroimaging: Brain CT, MRI (cerebral palsy)
- Lab and other ancillary tests: co-morbid allergy, immune dysfunction, GI disorders, sleep disorders, anemia, lead exposure, thyroid function, glucose and amino acid metabolism.

# Clinical diagnosis of ASD

- Diagnosis depends on the presence of core deficits, but there is tremendous variability amongst individual patients on the spectrum.
- Discrete Endophenotypes (ASD-subcategories based on common clinical features) with genetic markers may emerge.
- Personal approach: define strengths and challenges to develop a treatment plan to help individuals meet their potential.
- Resilience/Independence
- Have expectations

# How do you treat Autism?

- Treatment is highly individualized
- “If you have seen one autistic patient, then you have seen one autistic patient” (little generalization from one patient to another)
- Primary goal for me is to characterize specific strengths and deficits rather than fixate on issues of diagnosis.

# **Conventional Therapeutic Interventions –Multidisciplinary Approach**

- Educational programming
- Behavioral Therapy (ABA or Lovaas)
- Floortime or DIR-model (Greenspan)
- RDI, Pivotal Response
- Physical therapy
- Occupational /Sensory Integration Therapy
- Speech and language therapy
- Social Skills
- Pharmacologic (medications)

# Pharmacologic Treatments

- There is NO PHARMACOLOGICAL CURE for Autism.
- Purpose is symptom modification to allow child to participate in therapy/school
- Target specific therapy-limiting behaviors (self-injurious behaviors, aggression, hyperactivity, impulsivity, stereotypies, rigidity/perseveration, oppositional and anxiety).

# Drugs for “better behavior”

- Restricted interests/repetitive behaviors
  - SSRIs: fluoxetine\*, paroxetine, sertraline, citalopram
- Aggression or Self-Injurious Behavior
  - Atypical neuroleptics: risperidone\*, aripiprazole, olanzapine, quetiapine
  - Anti-convulsants: carbamazepine, valproate\*, lamotrigine
- Attention
  - Stimulants: methylphenidate\*, dextroamphetamine
  - Non-stimulants: atomoxetine
  - Alpha-2 agonists: guanfacine, clonidine
- Cognition
  - donepezil (Aricept)

# Complementary and Alternative Medicine (CAM) Interventions

- Research suggests that between 30 and 95 percent of children with ASD have been provided with complementary or alternative medical treatment. However, less studied.
- GF/CF diet, Biomedical intervention e.g. vitamins, supplements, omegas
- Detoxification or “chelation therapy”
- Auditory Integration therapy
- Music Therapy/Neurologic Music Therapy
- Equestrian Therapy
- Hyperbaric Oxygen Therapy
- And Many More....



# Research Needs

- Needed for clinicians to validate and guide appropriate and individualized treatment
- My perspective, we need partnerships with community based practitioners and research centers
- Community based research looking at interventions that the *community* is using. (Animal therapy, music (NMT), OT/PT based interventions, cognitive programs, social interventions)
- Need to train community physicians about research
- Research needs to be easily translated
- Research for needs of our transitioning young adults.  
**HUGE NEED**

# ALTERNATIVE APPROACHES to rational intervention in ASD

- We know that early behavioral identification and intervention “works”. In fact the use of drugs is largely to make such treatment more effective.
- Early intervention works by taking advantage of neuroplasticity (whereby other areas or networks in the brain take over for aberrant or lost function).
- ASD has frequently been described as a disorder of “abnormal connectivity” between brain regions.

# Autism: The Abnormal Connectivity Hypothesis

- Local/Regional neurofiber connections are overabundant, but long inter-connective processes between different regions of the brain are reduced. The brain has difficulty in communication with itself.
- This results in abnormal processing of sensory stimuli, abnormal motor output, which leads to the behavioral manifestations associated with autism.

# ALTERNATIVE APPROACHES to rational intervention in ASD

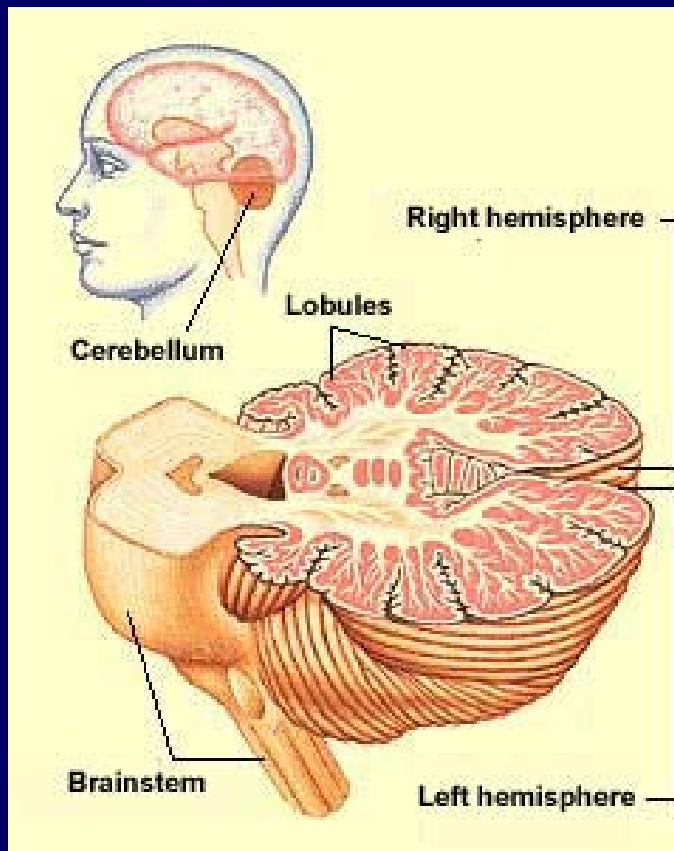
- This got me thinking about the foundations of brain development and how skills are developed
- Based on my practice philosophy to identify individuals strengths and challenges, I observed that as my patients entered school age, they would have continued difficulties with motor coordination, planning, postural tone, bilateral integration (socially awkward and clumsy)
- Biologic basis of this??
- Known consistent pathologic finding regarding loss of Purkinje fibers in cerebellum
- Is it possible to rationally intervene in a long-recognized and well-known COMMON anatomical abnormality associated with ASD in order to enable or restore neuroplasticity towards more “typical” development. Improve motor coordination and social interactions??
- Clinical phenotype??
- I believe it is possible.....

# Hypothesis for consideration

- I started clinically observing and documenting changes
- We have hypothesized that implementing a physical rehabilitation program that seeks to enhance automaticity to overcome developmental motor incoordination (clumsiness) can significantly impact changes in the cognitive domain (as dynamically assessed by instruments measuring IQ and executive function).

# The cerebellum in ASD

**The cerebellum in ASD has loss of Purkinje Cells.**



**The Cerebellum regulates automaticity (e.g.: “muscle memory”).**

- Cerebellar dysfunction can impede development of motor skills, as well as, executive and cognition function.
- The cerebellum streamlines implicit and explicit (declarative) learning and memory.
- The cerebellum is also important for communicating language and our intuitive understanding of emotions.

# Rationale

- The cerebellum comprises only 10% of the brain's volume but contains over half of all the neurons in the brain.
- There is loss of PCs in ASD, and PCs are exceptionally vulnerable to environmental injury (e.g.: hypoxia).
- The cerebellum plays a critical role in visuo-spatial perception, auditory processing, verbal memory, sequencing, executive functions, and language. Key issues in ASD individuals

# Preliminary Data

- In a cohort of school aged ASD-kids, we have gathered preliminary data that suggest that a PHYSICAL rehabilitation program that was developed to overcome motor clumsiness actually yields measurable improvements in COGNITIVE (e.g.: IQ) component-measures.
- Since cognitive measures are critical to children with ASD-measures achieving Independent Living, improved social interactions, we believe that this observation needs to be further investigated.



# Follow-up

- We are presently trying to team up with UCLA and/or USC to both extend this observation, and to figure out the neural networks that are impacted by the rehabilitation.
- To be continued...

# Important considerations

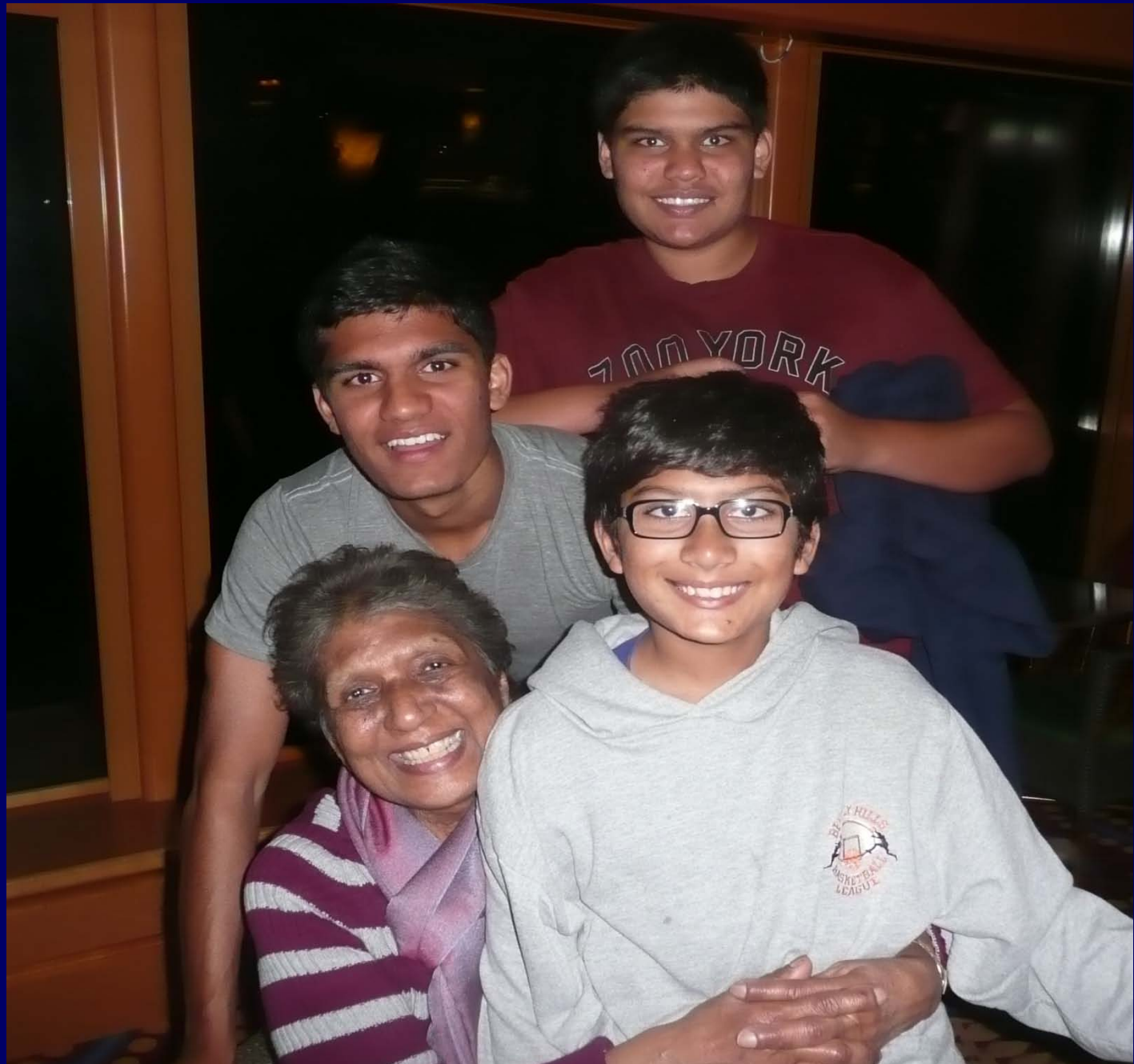
- **Most importantly, Listen to parents concerns early and throughout**
- **Early detection and intervention is key to improving and optimizing potential.**
- **Focus on strengths and challenges (affinities)**
- Resilience
- Independence skills
- Have expectations
- Knowledge is power

**My Hope...for ALL of Our Special  
Kids and Adults...**

**March to the beat of your own  
drummer as long as you march in  
the parade.**

Special thanks to  
My special kids  
and my mentor Dr. Audrey Griesbach





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