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The Brookings Institution
December 16,2005
Autism and Hope

Why Quirky?

- Affectionate
- Not pejorative or a euphemism
- Acceptable to many parents
- Inclusive of the wide range of ASD kids
- No prognostic significance
- Accurate

Features shared by ASD kids

- Developmental differences
  - Not always delays
- Temperamental extremes
- Sensory defensiveness
- Social difficulties
- Poorly developed Theory of Mind

Confusing Diagnostic Terminology

- Sensory Integration Dysfunction
- Pervasive Developmental Delay- NOS
- Pragmatic Language Disorder
- “Autistic Spectrum Disorder”
- Nonverbal Learning Disability
- Right hemispheric Dysfunction
- Asperger’s Syndrome
- High Functioning Autism

What does this mean for families?

- Doesn’t follow “What to Expect” guidelines
- Enormous stress of raising an atypically developing child
- Language delays/differences>>frustration>>behavior problems
- School difficulties
- Social difficulties
- Occupational difficulties
- Longer term dependence on parents

What does this mean for our communities?

- Increasing numbers of children with developmental disabilities
- Huge implications for school districts
  - ASD children often need specialized services and educational setting
  - Expensive and labor intensive
  - Huge discrepancies between communities

Prevalence

- Current data suggest that 1/166 kids in the US have an ASD diagnosis
- 4 times more prevalent in boys
- Siblings of affected children are at higher risk, so many families with multiple ASD kids.

What the data shows

- Estimates of children with developmental disorders range from 5-16% of children < 5 years of age
- 1/166 children with ASD
- California: number of cases of autism doubled between 1998 and 2002
- 1/25 households with a disabled preschooler

More data…

- 2004 average age of diagnosis of ASD 3-4 years
- Signs in most children are present by 18 months
- Cohort of children who seem to regress
  - 25%
  - First birthday videos subtle abnormalities
- Baby sibs project attempting to identify *very* early signs
- Earlier referral→earlier intervention→ better outcome

Is there truly an increase?

- YES!
- Also better assessment and recognition
- Shift in terminology has an impact
  - Some kids previously diagnosed as MR
- Trend toward parental diagnosis after children are diagnosed
- Various theories: genetics, assisted reproduction, toxins in environment, geographic hotspots
- Genetics: 90% heritable, 6-8% recurrence in sibs

Why pediatricians matter

- Most children receive primary health care from a pediatrician, nurse practitioner, or family practitioner
- A typical child has an average of 11 encounters with a health care provider in the first 2 years of life
- Great opportunity to observe development, temperament, parent-child interaction *over time*
- In the grand scheme, pediatricians are *not* the most important people in a quirky child's life!

Pediatric Medical Providers

- Gateway to services for the vast majority of kids
- Families get information and advice from their pediatricians
- Families may be inappropriately reassured that child's development is normal
- Most insurers require a referral for a specialty evaluation

Children with Disabilities

- Twice as many encounters with pediatric care providers
- Only one third to one half are identified prior to kindergarden entrance
- Failure to recognize?
- Failure to refer?
- *Failure to listen?*

What parents say

- “Throughout Abby’s first year, each time I talked with my pediatrician he assured me everything was fine…In the pediatrician’s office at the age of two, something set her off and she started tantrumming. She was fearful and overwhelmed and just couldn’t stop. The pediatrician said, “I would just ignore all this attention-seeking behavior.” He told me he thought I was decompensating and referred my husband and me for counseling.”

What parents say…

- “Aidan’s pediatrician felt he was within the norm. He became oppositional as soon as he could talk, and he talked very early. His tantrums simply couldn’t be ignored. He would scream and hang on to me, save his worst behavior for me. Again the pediatrician felt this was the usual oppositional/control issues of a toddler—and his recommendations didn’t help. It was impossible to do time out with Aidan. You would have to hold him down the whole time, which defeats the purpose of the time-out.”

Implications

- Failure to refer delays onset of diagnosis and services
- Excellent data to support Early Intervention
- Delay in one domain will have an impact on development in other domains
- Skills build on one another
- Parents questioning themselves and are questioned by family members

- “Compensating for missed opportunities, such as the failure to detect early difficulties…often requires extensive intervention, if not heroic efforts, later in life.”
  - National Academy of Sciences, *From Neurons to Neighborhoods*

Race and ethnic implications

- White children tend to be identified earlier
- Pennsylvania study of Medicaid population:
  - White children were identified with ASD more than a year earlier than their AA or Latino counterparts: age 6.3 years for white children, 7.9 for AA children, and 7.4 years for Latino children
- 6.3 years is *not* an early diagnosis

Improved outcomes with EI

- Immediate and long term benefits include
  - Better intellectual, social and adaptive behavior
  - Less special education services
  - Less anxiety
  - Increased HS graduation rate
  - Better employment rates
  - Decreased criminality
  - Decreased teen pregnancy rate

Even better outcomes

- Earlier the intervention begins, more profound the improvement
- Identification can begin in the first year of life
- *Intensive* early intervention associated with best outcomes over time

  Floor time

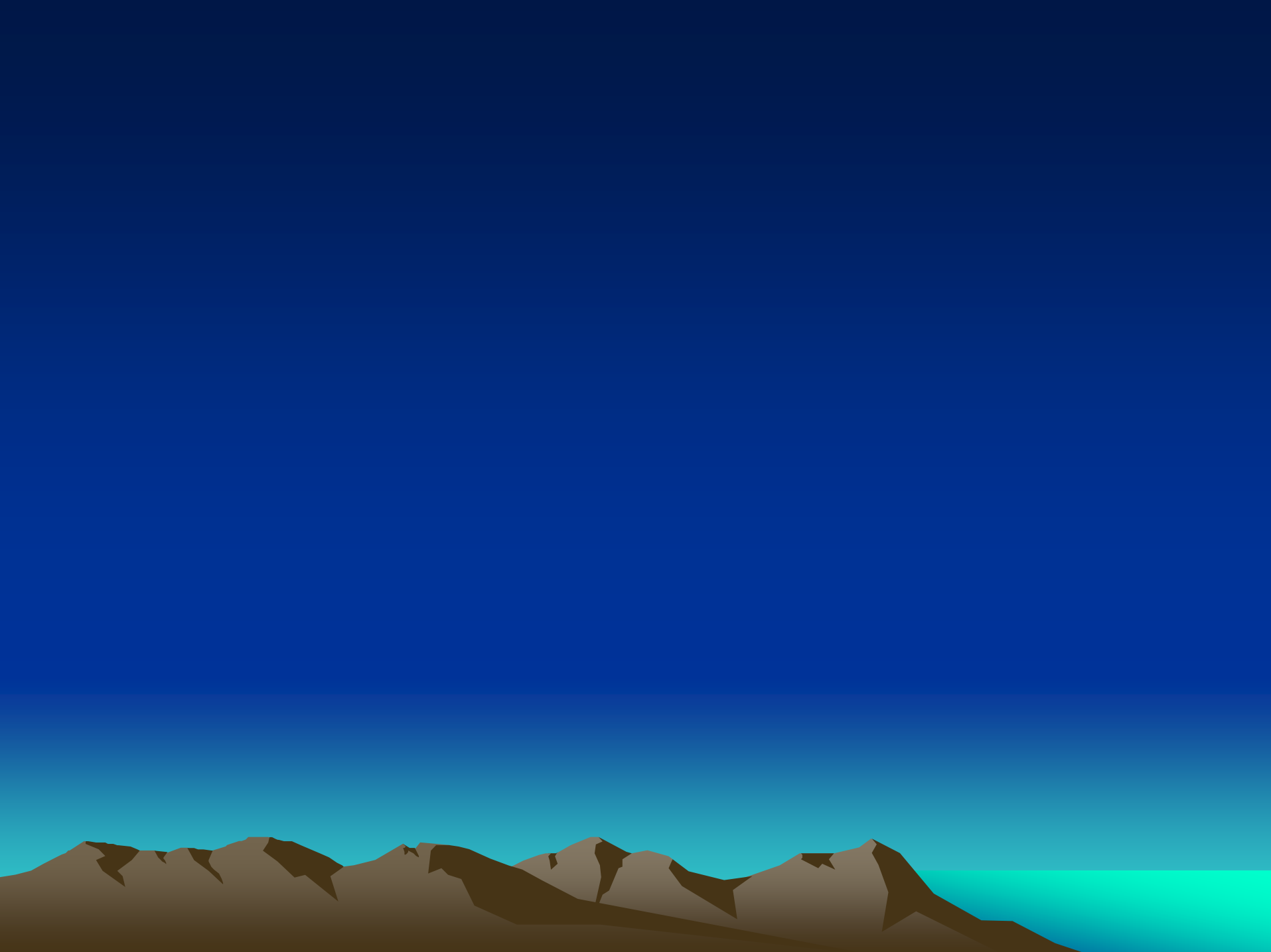
  ABA
- Some older children “no longer on the spectrum”

Challenge for pediatric practice

- Shift in the nature of pediatric practice just as prevalence of ASD is on the rise
  - Fewer infections, more development
- Increase in parental concern about behavior and development
  - 1958: 2% parents' concerns
  - 1960's: 45% more concerned about behavior than other issues
  - 1984: 81% questions concerned psychosocial issues.

Current practice

- Fewer than half of families report developmental screening by pediatricians
- Few than a third of pediatricians report screening
- Parents of children who are screened are more satisfied with the care they receive
- Most pediatricians report using informal methods such as checklists on encounter forms which are not validated
- Training hasn’t caught up with recommendations, older pediatricians not informed



On The Other Hand…

- Lack of parental concern does not imply normal or typical development!
- Pediatricians report expressing concerns to parents
- Many developmental concerns are raised only once, and by return visit are no longer an issue

Associated concerns

- No script for a given diagnosis
- Families are left to design their own program
- Gap in access
- Third party payors reluctant to cover services
- The immunization debacle
- Very little data to depend on
- Many alternative therapies, some dangerous: dietary interventions, vitamins, chelation, treatment for undocumented infections

Huge array of therapies

- Intensive Early Intervention
- Occupational Therapy/Sensory Integration
- Speech and Language/Pragmatics
- Physical Therapy/ Adaptive PE
- Friendship/Social Skills groups
- Individual psychotherapy
- Medications

Immunizations

- Primary responsibility of pediatricians
- Andrew Wakefield paper in Lancet suggesting and association between MMR vaccine and autism
- Paper subsequently retracted
- New fear is thimerisol—no conclusive data, removed from vaccines
- Major public health concern due to increase in unvaccinated children

AAP Autism Expert Panel A.L.A.R.M.

- Autism is prevalent
- Listen to parents
- Act early
- Refer
- Monitor

Obstacles to screening

- Time and cost constraints
- Need for detailed documentation
- Reimbursement
- Cultural and language barriers
- Lack of interest, knowledge, or training
- Lack of awareness of community resources
- 81% parent questions re: development and behavior—not all reflect a true disability

Red flags for referral

- No babbling, pointing or gesturing by 12 months
- No single words by 16 months
- No two-word spontaneous phrases by 24 months
- Any regression in language of social skills at any age

Higher Risk Populations

- Siblings of ASD children (4-7%)
- Maternal drug/alcohol abuse, acute /chronic illness
- Prematurity, low birth weight, multiples
- Intrauterine growth retardation
- Infection (sepsis or meningitis)
- Poor feeders or growers
- Micro/macrocephaly
- Internationally adopted children

What happens to quirky kids?

- Difficult to prognosticate
- Huge range
- World is more tolerant of quirky adults
- Some children will “outgrow” much of their quirkiness
- Not all are college bound

Looking forward

- We are a much richer world because of the quirky people among us.
- We have a responsibility to nurture children with developmental disabilities to reach their greatest potential.
- We need more data??
  - National Children's Study

- There truly is HOPE!

What is a pediatrician to do?

- Plan how and when to screen in your particular practice setting
  - Who will administer the instrument?
  - What instrument will be used?
  - How long will it take?
  - At what age will it start?
  - Are norms available for this population?
  - Other languages?
  - Less educated parents?

Fine motor adaptive delay

- Isolated or combined with other delays
- Visually impaired or cognitive impairment such as mental retardation
- Older preschool or elementary children may represent coordination disorder or dysgraphia
- Often in association with ADHD/LD

Parental concerns

- Expressive language delay most common developmental presentation in primary care
- Usually comes up when child is > age 2
  - May be an indicator of hearing loss, mental retardation, autism, dysarthria, learning disability, or developmental language disorder
  - May be none of the above (most likely)
  - Helps to have a trained eye, family history, other aspects of behavior and development

When parents are concerned about hearing

- Listen to the nature of the complaint
- Less common but more serious concern
- Universal hearing screening in the newborn nursery makes a later diagnosis less likely
- Atypical hx red flag: inconsistent symptoms
  - Doesn’t respond to his name but hears environmental sounds well
  - Seems to be in his own world, poor eye contact.
  - Doesn’t seem to notice mom entering or leaving room

Parental concerns

- Gross Motor Delay
  - Great concern to parents of children 6-18 months of age
  - Important to distinguish if isolated motor delay or combined with other delays
  - Older children may have coordination disorder where motor skills lag behind other skills
    - ADHD, Asperger's, LD

Personal or Social Delay

- Is the environment one of deprivation, abuse, or neglect?
- Reactive attachment disorder- seen in older adopted children.
- *Impaired joint attention - unique and almost universal in ASD*
  - Typically emerges between 6 and 14 months
  - Following a caregiver's gaze, a point, showing and pointing

Social-emotional problems

- Quality early relationships can positively effect neural connections and the organization of the brain, with lifelong positive effects
- Social and emotional problems resistant to change once established
- Associated costs are huge!
- EI provides quality early relationships!

Early signs

- Absence of appropriate development rather than presence of something
- 6-12 months tend to be quiet, not much babbling, not very social
- 3-9months: the “still face” paradigm—typical infants will find an expressionless face very disturbing, will cry, spit up, look away
- 14-18 months deficits in pretend play
- 18-30 months lack of reciprocal interaction, lack of a warm and joyful gaze

Approaches to screening

- Clinician questions-must be consistently asked at every visit
- Questionnaires -- Ages and Stages
  - Covers communication, gross motor, fine motor,problem solving,personal-social, over 5 years
- PEDS-Parents Evaluation of Developmental Status
- CHAT-Checklist for Autism in Toddlers

CHAT (18 months)

- Parents questions:does your child
  - Enjoy being swung, bounced on your knee,etc?
  - Take and interest in other children?
  - Like climbing on things, such as stairs?
  - Enjoy playing peek-a-boo/hide-and-seek?
  - Ever PRETEND, for ex, to make a cup of tea using a toy cup and teapot or other pretend things?
  - Ever use his/her index finger to point, to ask for something?
  - Ever use his/her index finger to point to indicate interest, that is to get you to look at it?
  - play properly with small toys (cars or bricks)
  - Ever bring objects over to you to SHOW you something?

CHAT--Observation

- During the appt, does the child make eye contact with you?
- Get child's attention, then point across the room at an interesting object and say, "Oh look! There's a balloon!" Watch child's face. Does the child look to see what you are pointing at? To record YES, ensure the child did not look at your hand, but actually looked at the object to which you pointed.
- Get child's attention, then give child a miniature toy cup and teapot/pitcher and say, "Can you make a cup of tea?" Does the child pretend to pour tea. If you can elicit an example of pretending in some other game,

CHAT-Observation

- Say to the child, “Where’s the light?,” or “Show me the light.” Does the child point with his/her index finger at the light? Repeat with, “Where’s the door?” if child does not understand “light.” To record YES, child must also look around as s/he points.
- Can the child build a tower of bricks (how many?)

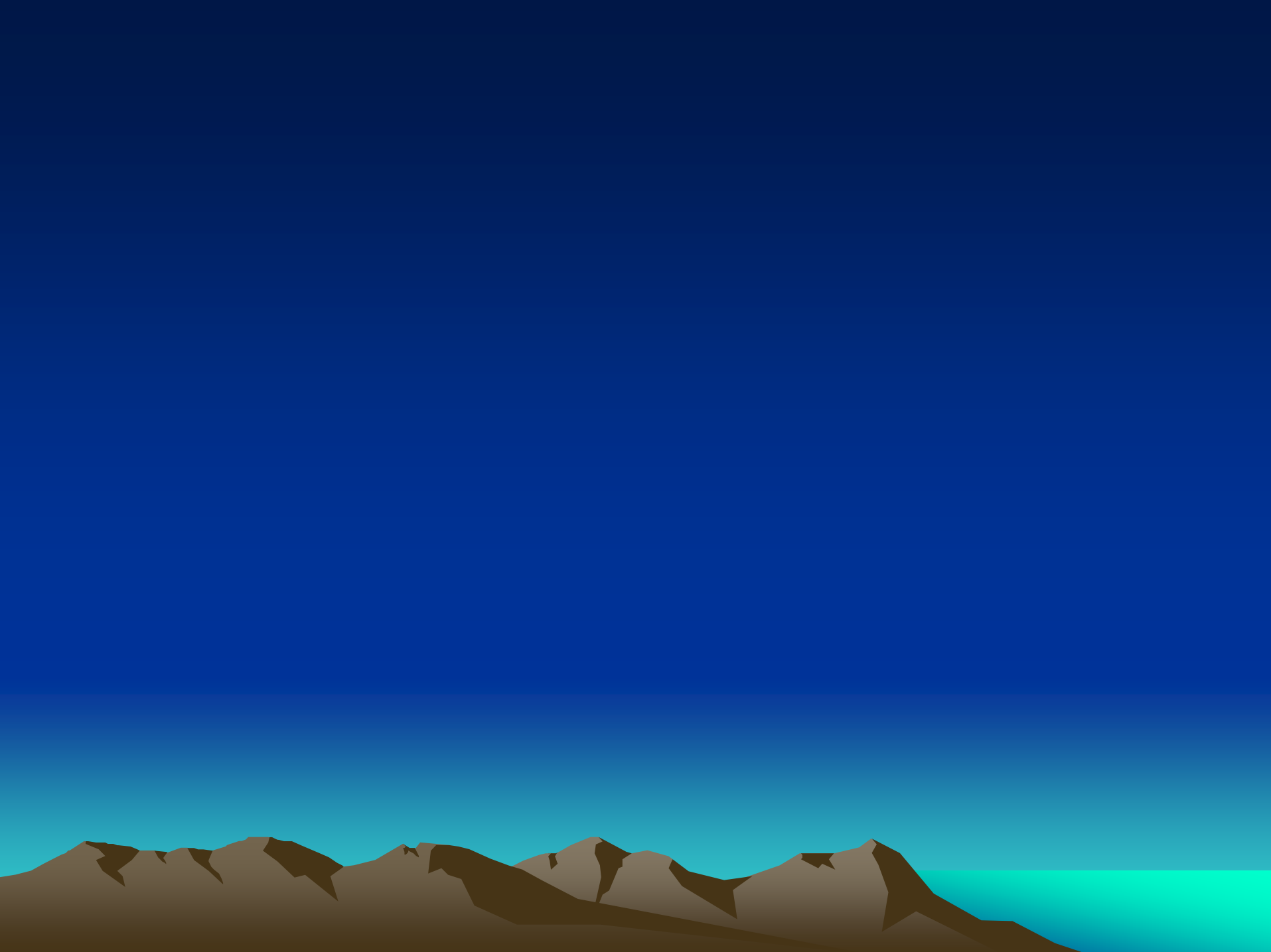
Higher Risk Populations

- Lead or other toxic exposures
- Neonatal seizures
- Congenital malformations
- Internationally adopted children
- Adolescent or single parents
- Mentally ill or retarded caregiver
- Inadequate resources/poverty

The earlier signs

What are parents to do?

- Repeat concerns-be a squeaky wheel
- Don’t wait for next well-child visit!
- Talk with daycare or preschool teachers
- Bring in a note from teacher
- Talk with school nurse
- Make a separate appointment
- Self-refer to Early Intervention
- Talk with your insurance company



- First signs (firstsigns.org) excellent website for parents and professional on early detection

Four domains of development

- Communication
- Motor
- Cognition
- Affective/social-emotional

Pediatric Screening Guidelines

- AAP Committee on Children with Disabilities recommends a developmental screen at each well-child visit
- AAN/CNS joint practice parameter
- Screening vs. surveillance
- Include the parents: experts on their own children
- Multiple screens over time most valuable

The ‘Tincture of Time’

- UK study: half of parents of children later diagnosed with ASD were reassured by their pediatrician and told not to worry
- Usual interval between parents’ first awareness and a definitive diagnosis of ASD in 4 years.

Current Practice

- Screening of the symptomatic child is not really screening
- “Screening” is for asymptomatic children
- Parent questionnaires are more accurate than non-standardized screening tests
- Mean interval between onset of parental concern and seeking professional help is 6 months:*by the time parents bring a concern to the office, 6 months of worry*

Obstacles

- A typical day in a pediatric office, a typical visit with multiple children, esp in winter!
- Children/parents anxious, esp if ill or shots
- Time or perception of time involved
- The doorknob moment…oh, by the way
- Parents hoping pediatrician will reassure
- Screening only when there is a problem