Diagnosis of Autism and Future Directions in Treatment and Research

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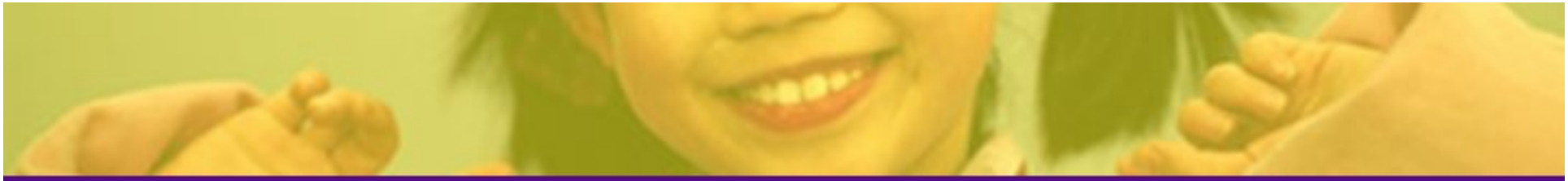
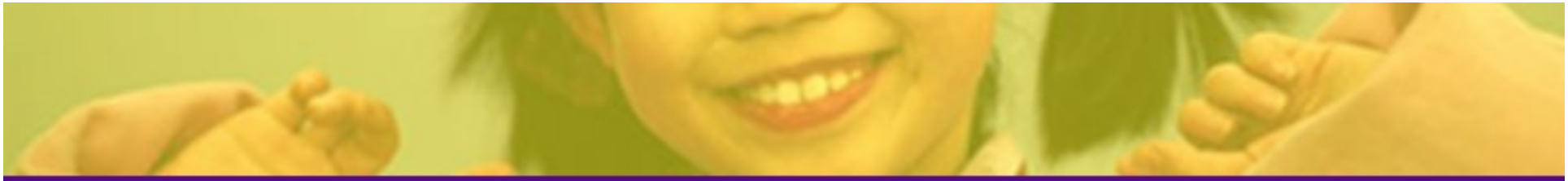


Diagnostics

- ASD – umbrella catch-all terminology
  - Dr. Kanner: Austrian-American, Johns Hopkins, 1943, psychiatrist
  - Dr. Asperger: University of Vienna, 1944
  - Heterogeneity and Idiosyncrasy (internationally)
  - Range of severity and limitations
  - Differential patterns of strengths and weaknesses
  - Complex presentations, no Polaroid snapshot diagnostic and treatment profile
  - 4:1 male-to-female ratio



MYTH

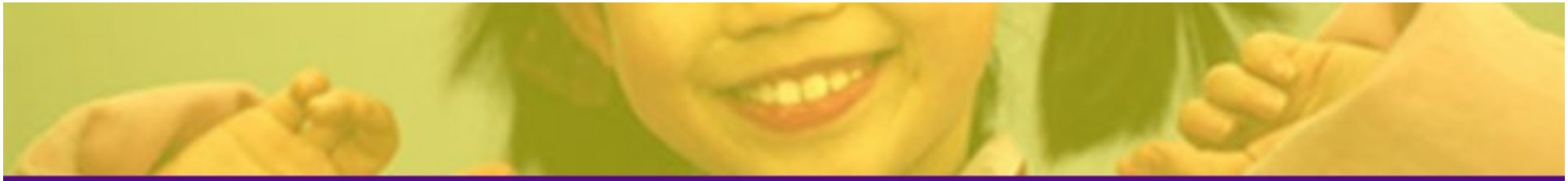
1. Aren't affectionate
2. Don't want friends
3. Have a "tic", it's OCD
4. Happens suddenly

5. Are mentally impaired (i.e., mental retardation)
6. No other disorders

7. Rare diagnosis
8. Psychiatric in type

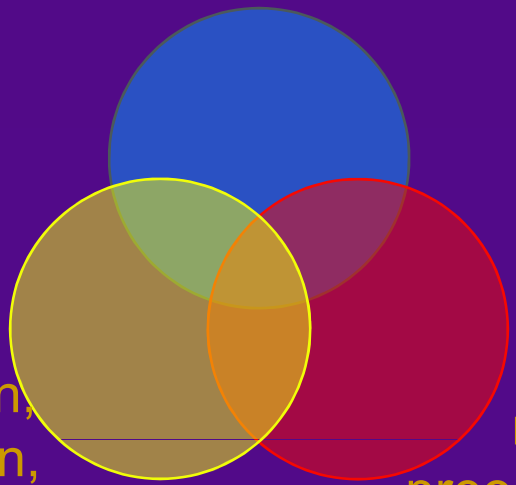
REALITY

1. On own terms
2. Difficulty w/social
3. Complex expression
4. Range- sudden, gradual, plateau, regress
5. Range of abilities, up to very superior
6. Often have comborbid dx, possibly higher risk
7. Est. range from 1:120 to 1:166, 1:154 for world
8. Neurodevelopmental, neurobiological



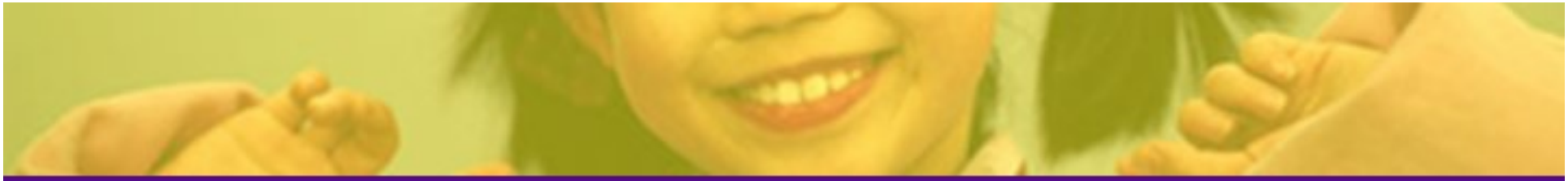
Triad of Impairment (what I look for in general)

Language, Communication (verbal & nonverbal)

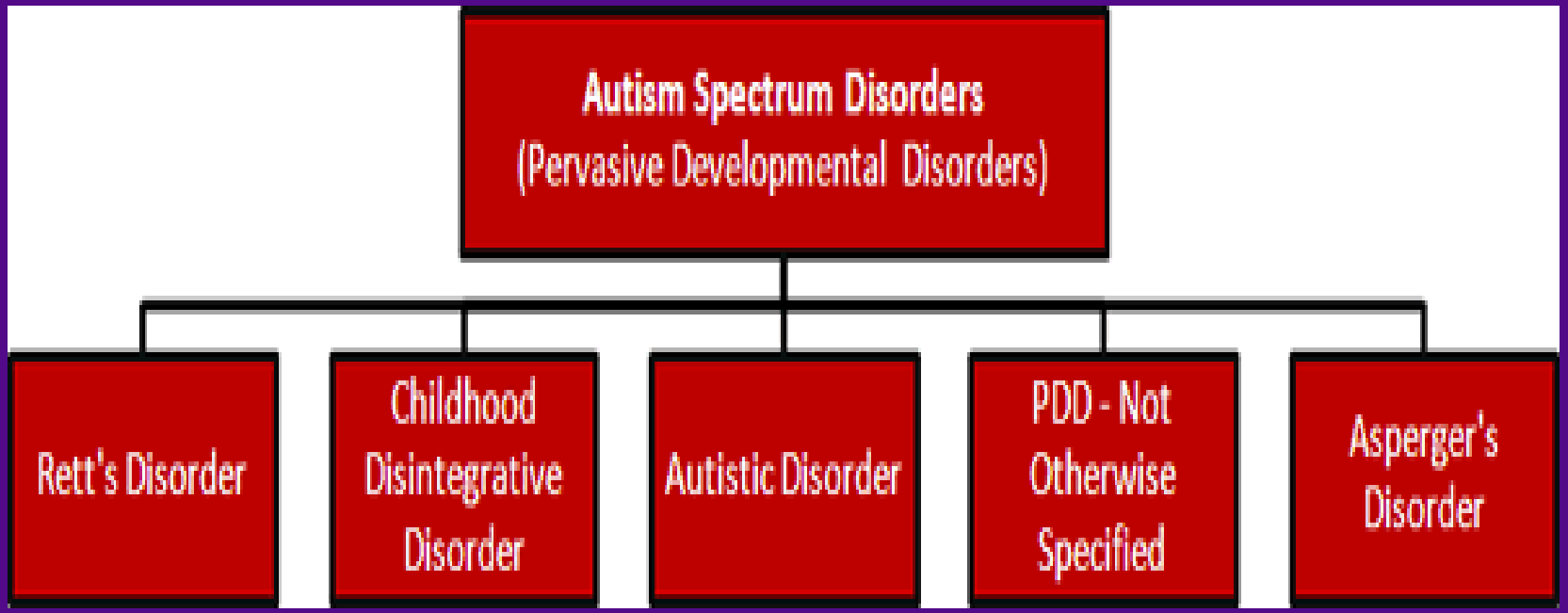


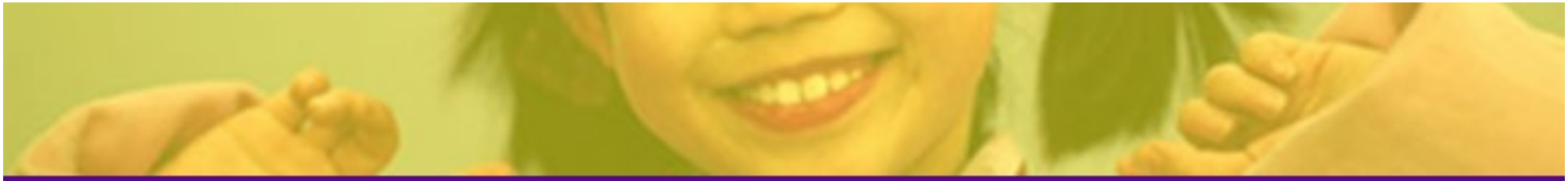
Social interaction, Communication, Emotional-behavioral regulation

Stereotyped behaviors, rigid interests and preoccupations (aka “insistence upon sameness”)



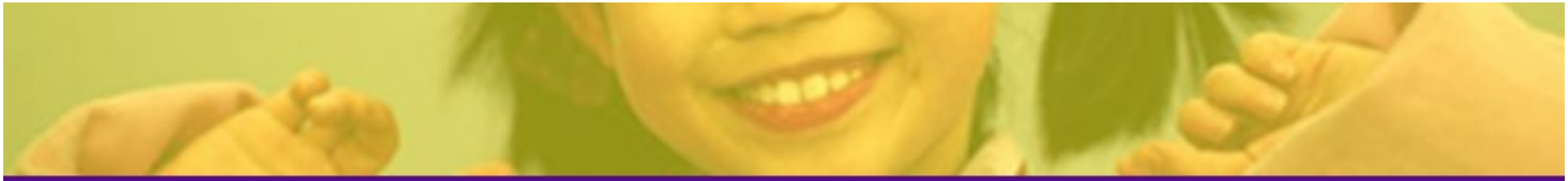
Disorders within the spectrum at present





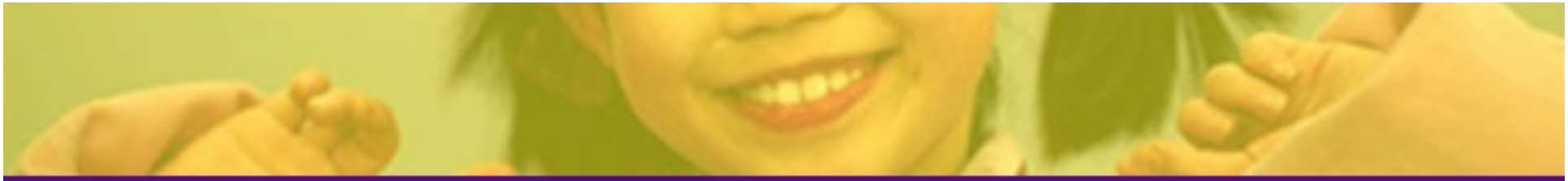
Diagnoses Within ASD

- Autistic Disorder – classic triad of impairment but still a range
  - Language delay, esp. for functional & social communication, pragmatics, significantly atypical speech (e.g., “Johnny-speak”)
  - Deficits in social engagement, interaction, and maintenance of play/communication activities
  - Display of stereotypies, rigid preoccupations and interests – COMPLEX
    - Stereotypies (e.g., hand-flapping) alone do not solely confirm of autism
    - Stereotypy vs. tics vs. OCD vs. self-stimming (and all combinations in between)
  - Often have sensory processing deficits – sensory seeking & aversions
  - May have largely age-appropriate motor function
  - ~ 70% - 75% with mental retardation (FSIQ ≤ 69)
    - Must be distinguished from bxs best explained under MR
    - Standardized assessment often difficult to complete with ASD
  - Early detection & treatment is KEY, typically by 18 months but currently much focus on abnormalities observable by ~10-12 months



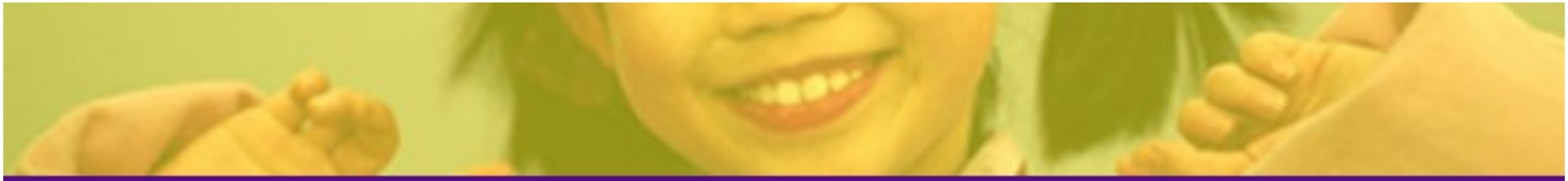
Diagnoses Within ASD (cont'd)
Asperger's Disorder/Syndrome

- Prominent deficits in social and emotional reciprocity, "theory of mind", "mind-blindness"
- "Higher end" of spectrum – controversial descriptors
- No significant delays in language, cognition, or adaptive skills (aside from social and independence), typically have above-average vocabulary
- Often have specific neurocognitive profile (not always)
  - Later onset (or observation) of deficits, + family history of Asperger's
  - IQ ≥ 110, VIQ > PIQ, pedantic (professor-like, fact-riddled) speech
  - Deficient fine motor, coordination & visuomotor integration skills
  - Phonetic spelling (lexical dysgraphia), difficulty with reading comprehension but may be hyperlexic
    - http://www.hyperlexia.org (controversial as an isolated syndrome, has been advocated for by some speech/language specialists across the yrs)
  - Memory – good rote simple, more deficient for more complex stimuli, initial encoding issues (maybe due to holistic reasoning deficits)
  - Asperger's vs. Nonverbal Learning Disability



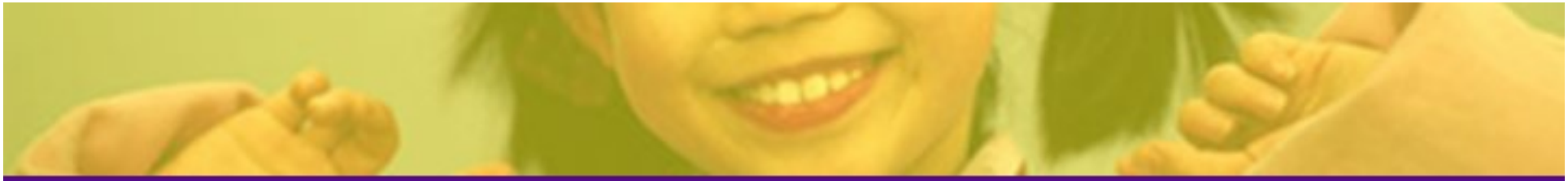
Rett's, CDD, PDD-NOS

- Rett's Disorder/Syndrome
  - Rare…1:10K to 1:15K, Typically female
  - Normal development until ASD appear ~ 6-18 mos
  - Underlying genetic mutation
- Childhood Disintegrative Disorder
  - Very rare…<2:100K, typically male
  - Normal develop until 3-4 y.o., ultimately more debilitated
- PDD-NOS/Autism Spectrum Disorder
  - "not otherwise specified", new NIH standards
  - Symptoms on the autism spectrum but doesn't meet criteria for a specific ASD



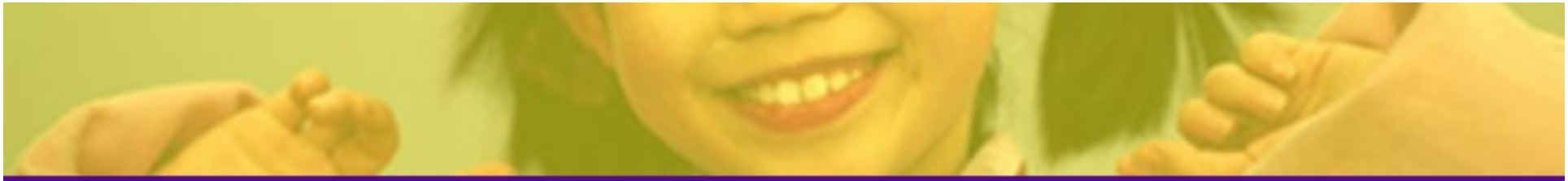
Diagnostic Considerations

- Early screening, pediatrician may administer the M-CHAT around age 16 to 18 months
  - 6 items pertaining to social relatedness and communication have best discriminability between children dx with & without autism/PDD
- Requires more than a 15-minute session, if concerns noted pt referred for comprehensive assessment
- Comprehensive multi-disciplinary evaluations
  - Physicians
    - Pediatrician/Developmental Pediatrician
    - Neurologist
    - Child Psychiatrist
  - Neuropsychologist, Developmental Clinical Psychologist
  - Evals & therapeutic support services with ST, OT, PT
  - Behavior analyst/therapist (BCBA preferable)
  - Genetics, allergist/immunologist, gastroenterologist, audiologist



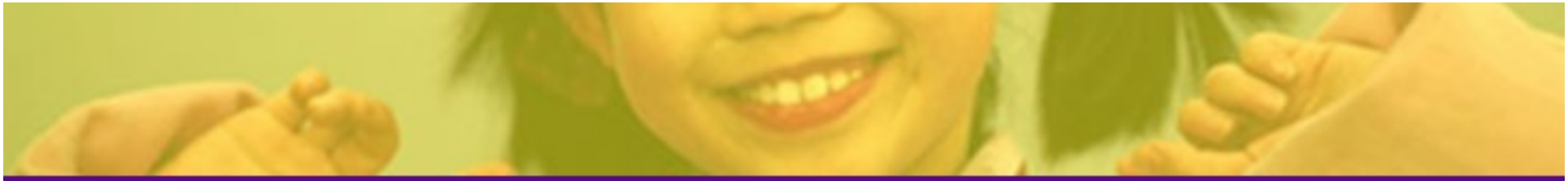
Treatments

- No isolated one-shot “cure” or “plan”
  - EARLY INTERVENTION
  - Ongoing evaluation and treatment, modify approach as goals are met
- Multi-method treatment approach
  - Pharmacological: typically treat most prominent symptoms first
    - Emotional-behavioral dysregulation
    - ADHD symptoms
    - anxiety common, but “anxiety” symptoms not always similar to anxiety disorders in general population re: triggers, response, etc.
    - SSRI, psychostimulant, AED, antipsychotic
    - Simvastatin being studied
  - Therapies
    - Speech/language, occupational (motor & sensory), physical
    - Applied Behavior Analysis, Floortime (Greenspan)
      - http://www.abaresources.com/
    - Formal social skills training with individual and group sessions (in-vivo training opportunities)



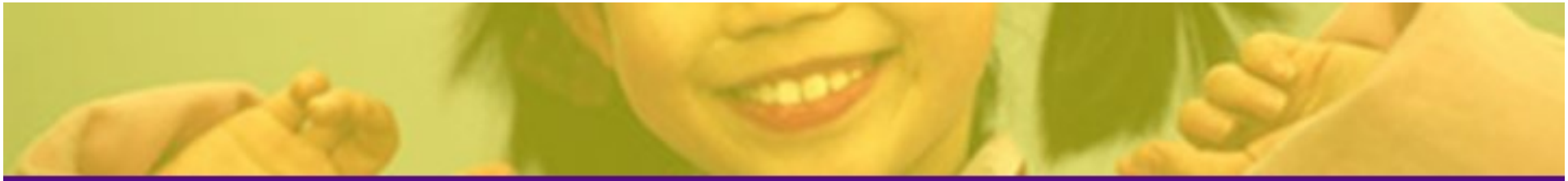
Treatments (cont'd)

- Diet, Supplements, Tests (DAN! doctors)
  - Gluten-free casein-free diet, "leaky gut" (may consult nutritionist)
  - Magnesium, B12, glutathione cream, cod liver oil, B6, zinc, MT protein (binds to mercury), chelation – removal of heavy metals, determine if overgrowth of yeast (hair, plasma, urine, stool analysis)
  - Hyperbaric oxygen treatments
  - Question if treatments are scientifically validated
- Academic - IEP, resource when needed, build strengths with typical peers, FBA with ABA therapist involved (REACH program in Shelby Co)
- Parent and Family Support, autism assistance dogs
- http://depts.washington.edu/dataproj/



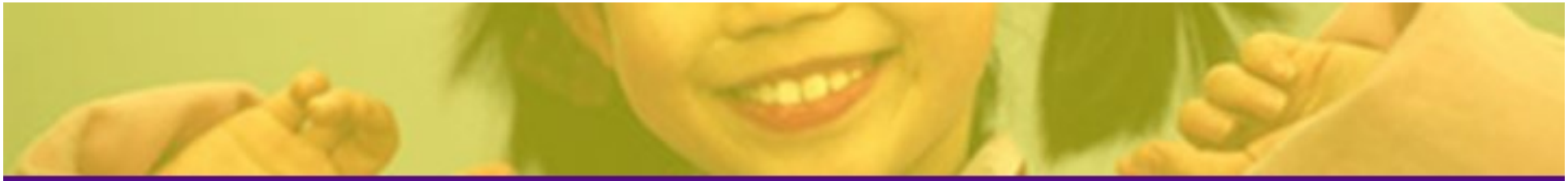
Scientific Research Advancements

- Main divisions of autism research
  - Identifying underlying neurodevelopmental factors
    - Genetics, phenotypic expressions, environmental contributions
    - Prenatal and perinatal factors under review
      - Hypoxia, advanced maternal & paternal age, birth weight, gestational age, maternal viral infection during pregnancy and subsequent maternal immune reaction (in animal models linked to impact on fetal development leading to behavioral symptoms of heightened anxiety & decreased social interaction)
  - Identifying features of ASD and developing diagnostic tools for early ID
    - checklists, standardized neurocognitive assessments, epidemiological studies, EEG, MEG, MRI, fMRI, SPECT, PET, audiological, etc.
  - Identifying and evaluating efficacious treatments for all of the problems within the spectrum
    - From speech/language to social to emotional-behavioral to pharmacological to school- and community-based initiatives, etc.



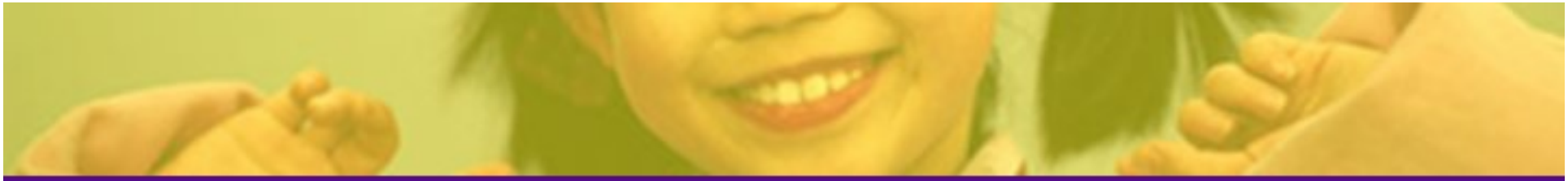
Scientific Research Advancements cont'd

- Genetics (genome scanning, genome-wide association tools)
  - Chromosomes are structures that contain genes
  - Rearrangements, deletions, duplications
  - Studies implicate "involvement of nearly every chromosome in the human genome", consistently replicated linkage findings on chromosome 7q, 2q,15q
  - Autism Genome Project
    - Recent from CHOP: kids w/ASDs more likely than controls to have gene variants on chromosome 5 in region located between 2 genes (*CDH9 & CDH10*) that carry codes to produce neuronal cell-adhesion molecules involved in neural communication, may contribute to up to 15% of ASD cases
    - Also at CHOP – copy number variations found are active on 2 gene networks that play critical roles in development of neuronal connectivity in the CNS
    - Harvard/Boston group – "hot spot" on 16p for ~1% of those studied
  - Twin Studies: Environmental influences important b/c concordance rates in monozygotic twins are < 100% & phenotypic expression of ASD symptoms varies widely (even within monozygotic twins)
    - Monozygotic twins - < 70% of twin pairs are concordant for autism, ~ 90% are concordant for a broader spectrum of related cognitive or social probs
  - Genotype and phenotypic expression, "complex inheritance" that may predispose/more susceptible



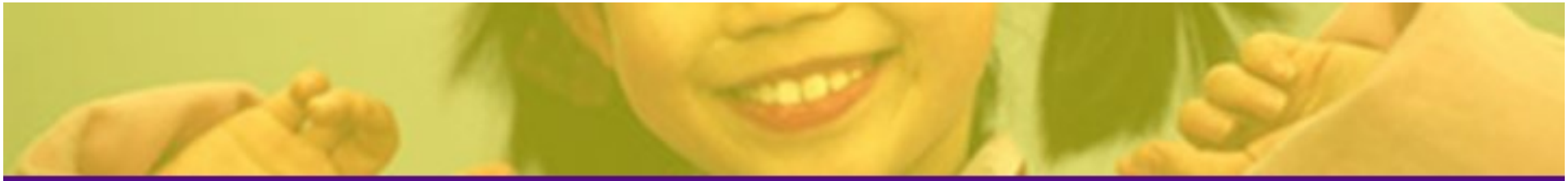
Scientific Research Advancements cont'd

- Neuroanatomy, neurobiology, neurochemistry
  - At present, no single region of the brain or pathophysiological mechanism identified in association with autism
    - Postmortem findings, animal models, & neuroimaging studies have focused on the cerebellum, frontal cortex, hippocampus, & amygdala. The cerebello-thalamo-cortical circuit may also be influential in autism.
  - White/gray matter differences, deficits in neural information pathways for processing, atypical large "growth spurts" in brain shortly after birth and then slower growth thereafter
  - Pericingulate sulci, left frontal, mesial temporal (including hippocami & limbic system), cerebellum
  - Altered serotonin metabolism
  - Reduction in cellular presence and communication
  - Auto-immune disorders/family hx, unusual cascade of immune responses in brain during sensitive periods of development
  - Macrocephaly due to neuroinflammation
  - < 2001 vaccines (thimerosol preservative is 50% mercury by weight)



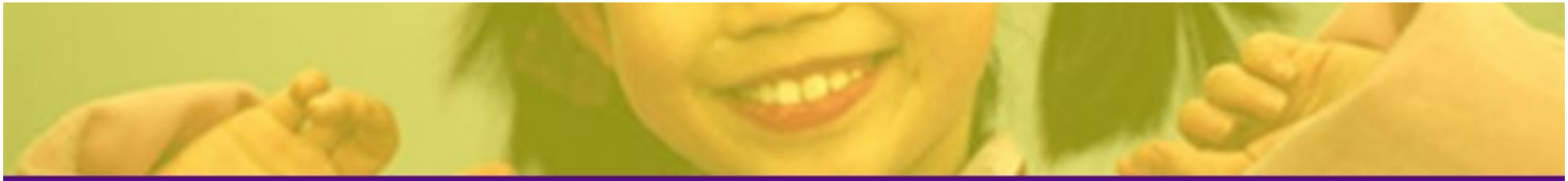
New Directions in Developmental Research for Diagnosing ASD

Early ID, 10-12 mos no or inconsistent response to name (early display of stereotypies in retrospect via home videos for some)	Language – functional needs & social (verbal/nonverbal)	Imaginative play 16 mos, mirroring emotional/social
Sensory seeking/ aversion, fail to explore as early as 18-20 mos	2–2½ yrs, no or limited involvement of/ with others	Active engagement in groups when exposed? (for some not known until preschool



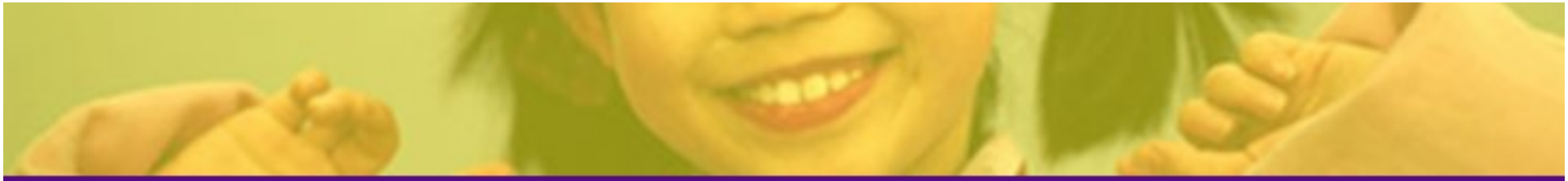
Subgrouping?

- ASD – multifactorial, polygenic etiology
- Current studies at Duke University involving psychosocial disorders in children with ASDs
- Journal of Child Neurology Jan 2008, prevalence of concurrent disorders in 160 ASD subjects
  - Sleep disorders 83 (52%)
  - Food intolerance 81 (51%)
  - Gastrointestinal dysfunction 94 (59%)
  - Epilepsy 22 (14%)
  - Mood disorder 42 (26%)
  - Aggression 51 (32%)
  - None 19 (12%)



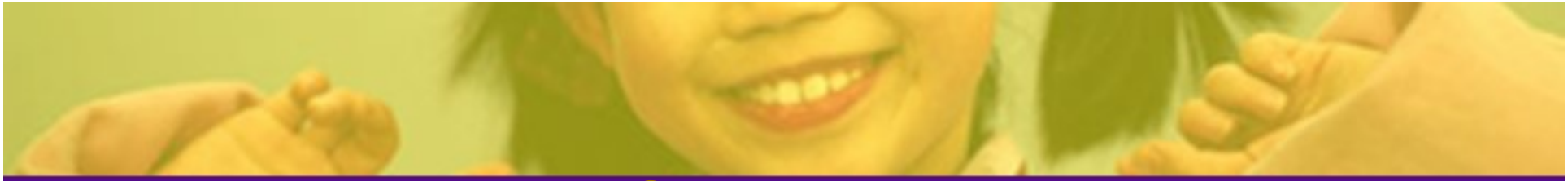
Community Resources

- TN Early Intervention, 901-937-6738
- North MS Early Intervention (Project Run, NMRC Oxford) http://www.msdh.state.ms.us/msdhsite/_static/19,0,166.html
- Center on Disabilities “Partners for Inclusive Communities” www.uams.edu/partners
- Le Bonheur Early Intervention & Development, 901-287-5858
- Harwood Dev. Center (18mos to 3yrs), www.harwoodcenter.org, 901-448-8369
- Transformations (ABA & school preK - HS) 901-647-9136
- www.autismsolutioncenter.org, 901-758-8288
- Midsouth ARC, 901-327-2473
- Autism Society of America, www.autism-society.org
- www.specialkidsandfamilies.org, 901-683-8787
- UT Boling Center for Developmental Disabilities www.utmem.edu/bcdd/
- Barbara K. Lipman Early Childhood School and Research Institute http://lipman.memphis.edu
- In Mississippi: www.teaam.org,1-866-993-2437
- Pediatric Specialty Care in AR, 870-733-1200
- Mid-South Autism Association (NE Arkansas area) 870-739-1366
- Autism Association of NE Arkansas, http://www.aanea.org/



Schools

- Transformations (901-647-9136)
- Hope Presbyterian Preschool inclusion class (3-5 y.o.) for special needs (901-844-HOPE)
- The Wesley School in Collierville – for special needs children K-7th grade (901-737-3762)
- Barbara K. Lipman Early Childhood School and Research Institute http://lipman.memphis.edu
- Memphis City Schools (exceptional children & health services) – Barbara Bolton 901-416-0203
- Shelby Co Schools (Div. Of Special Ed) – Jo Bellanti 901-321-2710



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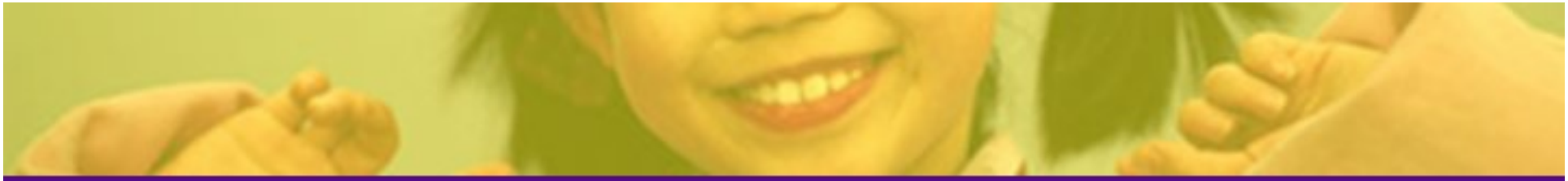

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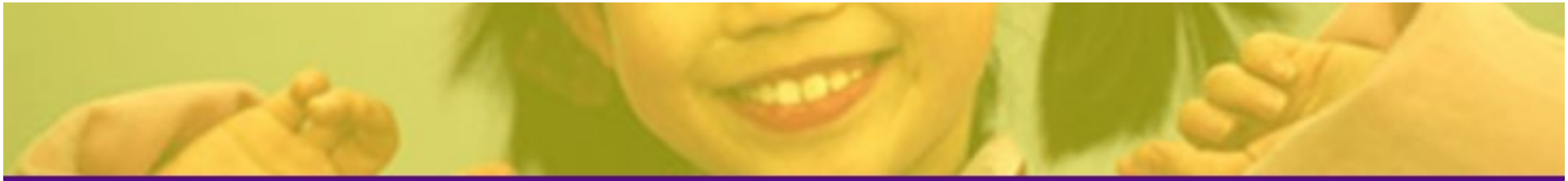

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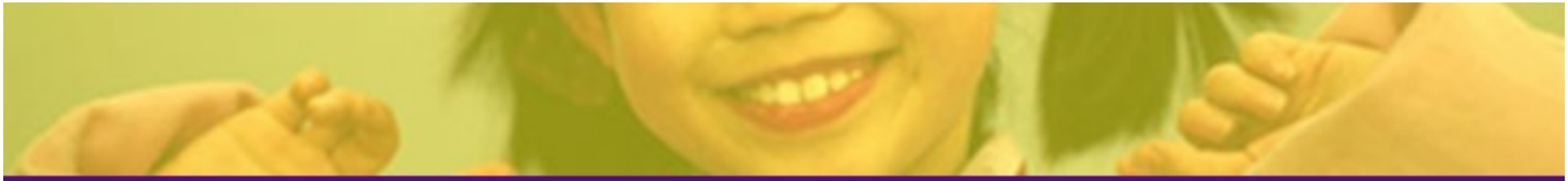

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