38TH ANNUAL CONFERENCE
OCTOBER 17-20, 2018
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SHERATON NEW ORLEANS HOTEL | NEW ORLEANS, LA

Pediatric Grand Rounds

Chair & Moderator

Philip Fastenau, PhD

University Hospitals Cleveland Med Ctr & Case Western Reserve Univ.

Discussants

Allyson G. Harrison, Ph.D., C. Psych., Queen's University

Karen Postal, Ph.D., Harvard Medical School & Lifespan Private Practice

Scott Hunter, Ph.D., University of Chicago Medicine

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Financial Disclosure

All presenters reported that they have no financial relationships to disclose.

National Academy of Neuropsychology, 2018

FETAL ALCOHOL SYNDROME IN THE CONTEXT OF MULTIPLE ETIOLOGICAL FACTORS

Presentation by Jack C. Lennon, MA

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Contributing Authors:

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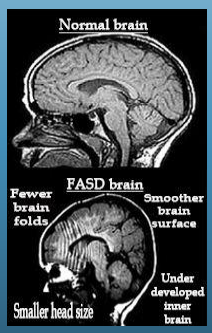
OBJECTIVES

- **Fetal Alcohol Spectrum Disorders**
  - Background
  - Diagnostic Protocols/Limitations
- **Case**
  - Referral
  - Demographics & History
  - Test Results
  - Diagnostic Impressions
  - Return to Protocols/Limitations

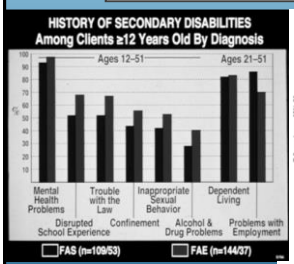


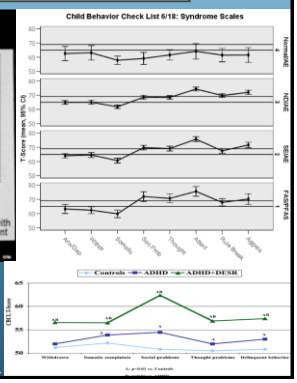
FASD BACKGROUND

- Difficult to diagnose accurately –estimates more common
  - 800-8,000 children born each year with FASD in US
  - FAS involved in 0.2-5.44 out of 1,000 live births in US
    - Global: up to 39.65/1,000 live births
  - 7.3% child-bearing-age women at risk for ETOH+ pregnancy
  - Prenatal ETOH exposure in 1% of live births
- Expected Neuropsychological Sequelae
  - Global delays
  - Pragmatic language
  - Emotional self-regulation
  - Attention
  - EF – problem-solving, planning, inhibition
    - Overlap with ADHD and other disorders

AAP; CDC; Burd; Glass,et al.; Greenbaum et al.; Peadon & Elliott; Roozen et al.

FASD BACKGROUND (CONT.)



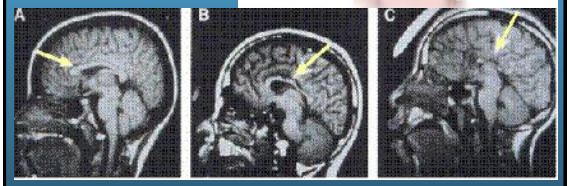


Brown et al., 2011; Streissguth et al; Edwards & Greenspan; Brown et al., 2010; Popova et al; Astley, 2010; Spencer et al.

NOFAS DIAGNOSTIC CRITERIA

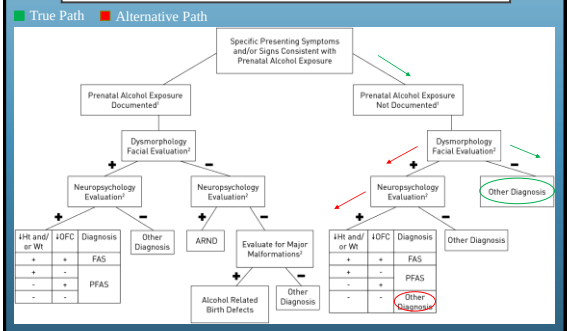
FAS:
1) Growth problems
2) Facial dysmorphia
3) CNS abnormality

Documented prenatal ETOH exposure is <u>not</u> necessary.



NOFAS; Center for Disease Control and Prevention; Thomas et al.; AAP; Marco et al.

AMERICAN ACADEMY OF PEDIATRICS



AAP; Hoyme et al.

REASON FOR REFERRAL

- Rule out Fetal Alcohol Syndrome (FAS) due to:
  - Mood dysregulation;
  - Legal history (ongoing);
  - Authority issues;
  - Impulsivity and inattention;
  - Behavioral concerns (aggression, HI, sexual behaviors, rule-breaking).

<u>NOTE: This is a neuropsychological referral for diagnostic/prognostic clarity, not a forensic evaluation pertaining to risk of recidivism or any other legal question.</u>

DEMOGRAPHICS/HISTORY

- 14-year-old Caucasian, heterosexual male;
- Right-hand-dominant;
- Adopted at ≈ 13 months;
- 8th grade (detention center schooling) w/ smooth grade transitions;
- Low-middle SES (bio mother low SES).

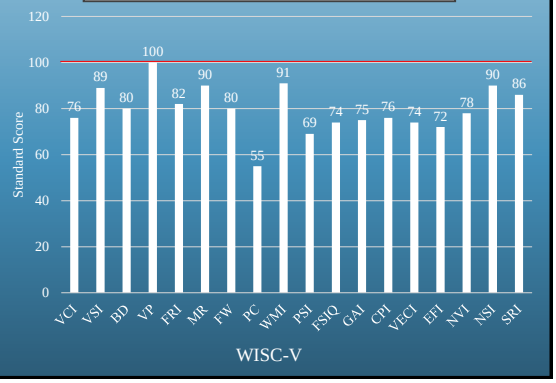
- Medical history
  - Preterm birth (29 weeks), asthma, otitis media, ADHD, Bipolar I Disorder, ODD, depression, insomnia
  - Average height/weight for chronological age
  - Denied hx seizures, known head trauma
  - No evidence of facial dysmorphia, microcephaly, + MRI findings, or past neuropsychological evaluation
- Bio M with history of polysubstance use (excluding ETOH)
- Bio M "threw" Pt into wall, per AM's report

HISTORICAL DATA (CONT.)

- Current medications
  - Aripiprazole 7.5mg bid
  - Divalproex 125mg tid
    - Reported inconsistent use
- **<u>Infant</u>**: frequently ill with respiratory issues
- AM reported walking at 11mo with regression at 13mo until 16mo
- **<u>Toddler</u>**: limited emotion
- **<u>Child</u>**: hyperactive, angry, aggressive with others
- **<u>Adolescent</u>**: angry, aggressive, authority issues, social with those who "don't get on [his] nerves," inhalant use 2-3 times
- Reported average academic performance
- Expressed HI toward 12yo brother (protective order filed by AM)

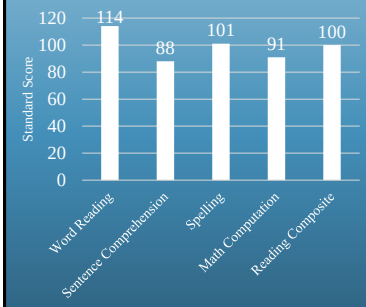
EFFORT/PERFORMANCE VALIDITY MEASURES					
<u>Rey Word Test</u>		<u>Rey 15-Item</u>		<u>Trails A & B</u>	
6	10	13		27s	74s
ADEQUATE		ADEQUATE		ADEQUATE	

INTELLECTUAL FUNCTIONING



WISC-V

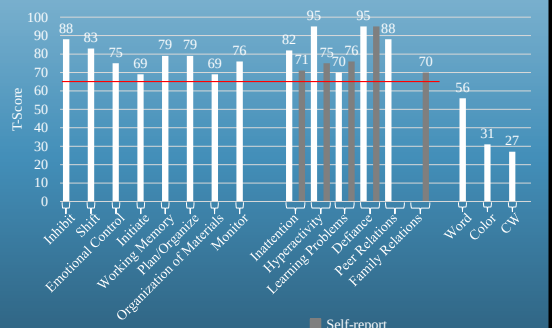
ACADEMIC ACHIEVEMENT



Domain	Grade Equivalent
Word Reading	11.4
Sentence Comprehension	6.0
Spelling	8.6
Math Computation	5.9

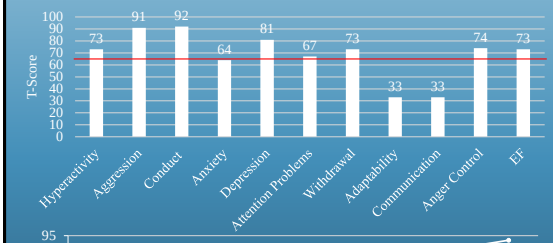
WRAT-4

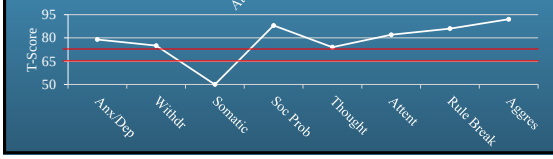
EXECUTIVE FUNCTIONING



BRIEF – Parent Form | Conners 3 | Stroop

BASC-3 & CBCL - PARENT



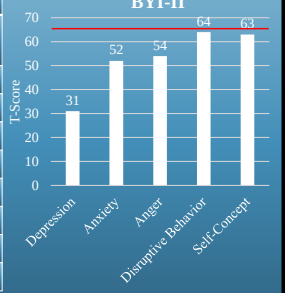


OTHER RELEVANT FINDINGS

ABAS-3 - Parent

Domain	Adaptive Level
Communication	Average
Academics	Average
Leisure	Ext. Low
Social	---
Community Use	Average
Home Living	Below Average
Health/Safety	Below Average
Self-care	Average

BYI-II



FASD BeST Met cutoff

ARND BCL Met cutoff

IMPRESSIONS & CONCLUSIONS

- F90.2 - Attention-Deficit/Hyperactivity Disorder, Combined Presentation, Moderate
- F88 - Other Specified Neurodevelopmental Disorder
  - *Characterized by:* Borderline IQ, EF deficits, developmental delay/regression, learning difficulties, possible exposure to prenatal toxins, possible early head trauma

R/O Major Depressive Disorder

R/O Posttraumatic Stress Disorder

1) More finely-tuned measures/profiles necessary

2) Determine need for documentation and facial features

3) Continue developing new treatments for disruptive bx in FASD

Mattson et al; Hoyme et al.; Ware et al; Coles et al.

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Neuropsychological functioning following methotrexate neurotoxicity stroke in an 11-year-old female with acute lymphoblastic leukemia (ALL)

Lindsay Katz, M.A.[1, 3], Madison Sunnquist, M.A.[2, 3], Ariana Garagozzo, B.S.[1, 3], Megan Scott, Ph.D.[3], & Scott J. Hunter, Ph.D.[3]
Roosevelt University[1], DePaul University[2], and
The University of Chicago Medicine[3]

Demographic Information

- 11-years-old
- Caucasian
- Female
- Middle SES

Reason for Referral

- Chronic headaches, acute widespread pain, blurry vision, and difficulties concentrating
- Decline in academic performance
- Social, emotional, and behavioral problems
- Completed comprehensive battery

Medical History

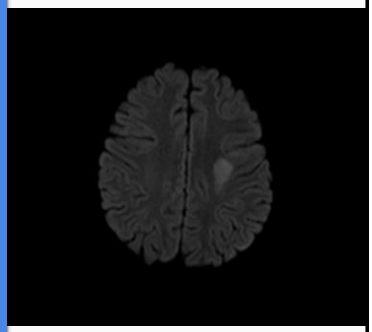
- January 2015: Acute lymphoblastic leukemia (ALL)
- July 2015: Left methotrexate (MTX) neurotoxicity stroke
- In remission since April 2017

Psychosocial History

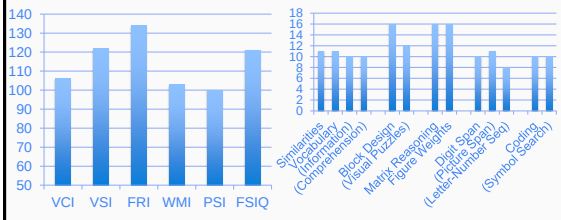
- Strong student
- Typical, age-appropriate friendships
- Older adopted brother with autism spectrum disorder (ASD) and fetal alcohol syndrome (FAS)

Imaging

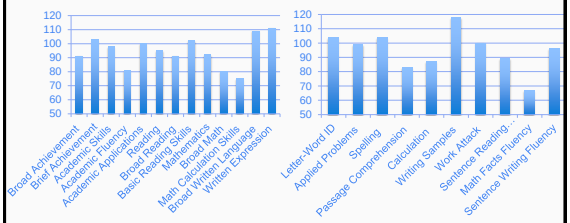
MRI
August 2015



Wechsler Intelligence Scale for Children, Fifth Edition



Woodcock-Johnson, Fourth Edition



Attention & Executive Functioning (EF)

- Low average performance on Auditory Attention of the NEPSY-II
- Overall average performance on other measures of attention and EF on the NEPSY-II and DKEFS; however, lower than would be expected given her cognitive abilities
- Immature and less efficient encoding strategies on the CVLT-C (serial clustering)
- Endorsed clinically significant concerns on the BRIEF-2 (self-report) with respect to her working memory and ability to shift between tasks

Motor and Visual-Spatial Functioning

- Overall average motor performance across tasks
- Stronger performance with her left, non-dominant hand on Grooved Pegboard and Fingertip Tapping on the NEPSY-II
- Average to superior visual-spatial performance, in contrast to reported vision difficulties

Social and Emotional Results

- Self and parent reported symptoms of anxiety, including panic, separation anxiety, generalized anxiety, and physical problems
- Self reported sense of inadequacy; parent report of symptoms of somatization and depression
- During the clinical interview, the patient's mother endorsed defiance and dishonesty

Impressions

- Superior intellectual abilities, with relative personal weaknesses in verbal comprehension and working memory
- Average academic skills, with low average to borderline impaired academic fluency
- Generally average objective attention and executive functioning, with immature learning strategies and subjective impairment in daily EF
- Average motor skills; slight non-dominant advantage
- Significant symptoms of anxiety leading to impairment across home, social, and academic environments

Thank you!

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Ariana Garagozzo, B.S.
Roosevelt University

Madison Sunnquist, M.S.
DePaul University

Megan Scott, Ph.D.
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Pediatric Grand Rounds: Case Study Cockayne Syndrome, Type III in 15-year-old female

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Cockayne Syndrome

- Prevalence is approximately 2.5 per million
- Neill-Dingwall Syndrome
- Autosomal recessive disease caused by mutations in either the *ERCC6* gene (*CSB*) or the *ERCC8* gene (*CSA*)
  - Type 1, "classic" or "moderate", diagnosed during early childhood
  - Type 2, "severe" or "early-onset", presenting with growth and developmental abnormalities at birth
  - Type 3, a milder form of the disorder

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Cockayne Syndrome

- Failure to thrive
- Cachectic dwarfism
- Photosensitivity (pigmentary retinopathy, cataracts)
- Sensorineural deafness and tooth decay
- Progeria
- Hypomyelination, calcifications and brain atrophy (supratenroial white matter, cerebellum, corpus callosum and brain stem)
- Neurocognitive deficits (memory, language, executive function dysfunction, social cognition, learning delays, visual-motor)

Case History

- Right-handed 15 year old female (DOB: 4/2018)
- Evaluation: 3/15/2018
- Referred by Neurology
  - Poor academic progress, loss of skills, short term memory problems, social skills, labile mood, anxiety, difficulty differentiating fantasy from reality, tremors, fatigue, hand and wrist pain

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Case History

- Delivered FT, 7lbs 5oz
- Developmental delays
- Brain MRI
  - demyelination pattern with abnormalities in cerebellar white matter extending from the periventricular zones to subcortical white matter
- Multiple therapies: OT, PT, Speech, psychotherapy
- 9th grade public school with IEP
- Lives with mother, step-father, and 2 siblings
- Fairly independent, mother considering quitting job
- Strained relationship with biological father
- 2010 neuropsychological evaluation, diagnosed with ADHD

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Neuropsychological Results: Effort, Orientation, Attention

Task	Raw Scores	Descriptors
WMS-III	14/14	Name, when/where born, president, day of the month, name of the place you are right now, what time is it
Reliable Digits	6	(passing ≤ 6) 3 forward, 3 backward with one round consistent and two rounds inconsistent
TOMM	48/50 50/50 50/50	

CPT-III:

- 90%ile or higher
  - Detectability
  - Commissions
  - Perseverations
  - Hit rate
  - Variability

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Neuropsychological Results: Intellectual Functioning

Wechsler Intelligence Scale, Children-5th Ed.

Index	Standard Score
Verbal Comprehension	86
Visual-Spatial Reasoning	84
Fluid Reasoning	85
Working Memory	65
Processing Speed	60
GAI	81

Subtest	Scaled Score
Similarities	8
Vocabulary	7
Block Design	6
Visual Puzzles	8
Matrix Reasoning	11
Figure Weights	4
Digit Span	3
Picture Span	4
Coding	1
Symbol Search	5

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Neuropsychological Results: Language

Developmental Neuropsychological Assessment, Second Edition (NEPSY-II)

Measure	Z-Score
Comprehension of Instructions	-4.9

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Neuropsychological Results: Memory

California Verbal Learning Test, Children's Ed. (CVLT-C)

Domain	Z-Score
List A Total Trials 1-5	-1.2
List A-Trial 1 Free Recall	-0.5
List A-Trial 5 Free Recall	-1.5
List B-Free Recall	-2.0

CVLT-C Cont'd

Domain	Z-Score
List A Short-Delay Free Recall	-1.5
List A Short-Delay Cued Recall	-1.5
List A Long-Delay Free Recall	-1.5
List A Long-Delay Cued Recall	-1.0
Recognition Hits	+0.5
Total Recall Discriminability	0.0

Neuropsychological Results: Memory

Wide Range Assessment of Memory and Learning, Second Edition (WRAML-II)

Measure	Z-Score
Sentence Memory	
Immediate Recall	-1.67
Picture Memory	
Recall	-1.33
Recognition	-1
Story Memory	
Immediate Recall	-1
Delay Recall	-1.33
Recognition	-0.33

Neuropsychological Results: Executive Function

Delis-Kaplan Executive Function System (DKEFS) - Verbal Fluency Test

Measure	Z-score
Letter Fluency	-0.66
Category Fluency	4
Category Switching Total Correct	3
Category Switching Total Switching Accuracy	5

DKEFS Cont'd

Measure	Z-score
Set-Loss Errors	13
Repetition Errors	10
Letter Fluency	8
Category Fluency	4
Category Switching Total Correct	3
Set-Loss Errors	13
Repetition Errors	10

Neuropsychological Results: Executive Function

Delis-Kaplan Executive Function System (DKEFS) – Color-Word Interference

Measure	Z-Score
Color Naming	-1.33
Word Reading	-1.67
Inhibition	-3
Inhibition/Switching	-1.67

Delis-Kaplan Executive Function System (DKEFS) – Sorting Test

Measure	Z-Score
Confirmed Correct Sorts	-1
Free Sorting Description	-1.33
Sort Recognition	-3

Neuropsychological Results: Motor/Visual Motor Integration

Finger Oscillation Test

Test	Z Score
Dominant (R)	-3.31
Non-Dominant (L)	-2.43

Grip Strength

Test	Z Score
Dominant (R)	-3.32
Non-Dominant (L)	-3.04

Grooved Pegboard

Test	Raw Score	Drops	Pegs	Z-Score
Dominant (R)	213	4	25	-14.12
Non-Dominant (L)	D/C	D/C	D/C	D/C

Neuropsychological Results: Motor/Visual-Motor Integration

Symbol Digit Modalities Test (SDMT)

Measure	Raw	Errors	Z-Score
Oral	35	1	-1.64

Benton Judgment of Line Orientation

Measure	Z-Score	Measure
Form H	-1.52	Form H

Beery-Buktenica Test of Visual Motor-Integration (Beery VMI) Sixth Edition

Measure	Raw Score	Z-Score
Visual Perception	26	-0.47

Neuropsychological Results: Academics

Wechsler Individual Achievement Test-3rd Edition

Subtest Cluster	Z-Score
Word Reading	0.87
Numerical Operations	-0.2
Spelling	0.27

Neuropsychological Results: Social Emotional

- Parent: Elevations anxiety, depression, somatic complaints, social problems, attention, executive dysfunction, low average adaptives
- Teacher: concerns with anxiety attention, executive dysfunction
- Self: Elevations with anxiety

*CBCL, BRIEF,Vanderbilt, SCARED, CDI-2, ABAS-3

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Summary

- Deficits related to executive dysfunction, attention, visual-motor integration, and receptive language
- Improvement noted when motor components were removed and verbal and visual reasoning abilities were low average
- Slow pace of learning not forgetful
- Academic performance surpassed estimated psychometric intelligence
- Diagnoses: Major Neurocognitive Disorder and GAD
- Recommendations: IEP with 18-21 year old programming, OT/PT/Speech, psychotherapy and wrap around services

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