Fetal Alcohol Spectrum Disorders

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Disclosure: Julian Davies, MD

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Overview

Define Fetal Alcohol Spectrum Disorders

Diagnosis

How the brain is impacted by alcohol

What to do about it

Who can help

What Is FAS?

Permanent birth defect syndrome caused by maternal alcohol consumption during pregnancy

An FAS diagnosis requires:

- Pre and/or postnatal **growth** deficiency
- Cluster of minor **face** anomalies
- **Brain** dysfunction
- Prenatal **alcohol** exposure

What about PFAS, AFAS, FAE, ARBD, ARND, etc?

Partial FAS, atypical FAS, fetal alcohol effect, alcohol-related birth defects, alcohol-related neurodevelopmental disorder, and all of the above without confirmed maternal alcohol exposure …

Patients missing one or more of the four FAS criteria

“FAE” has been retired; the alternatives aren’t much better

Fetal Alcohol Spectrum Disorders - an umbrella term, not a diagnosis

- We have a marker for FAS: “the face”
- We lack a specific neurobehavioral phenotype for FASD

A Brief History of FAS

The Current Problem

Leading known cause of mental retardation

- As common as Down Syndrome and spina bifida

Incidence of FAS in general population from 1-3/1,000 live births, similar in Europe

- Alaska Native/American Indians in Alaska: 5-6/1,000
- King County foster care: 10-15/1,000
- Russian specialized orphanage: 140/1,000

$17,000/yr in medical costs (9x those without)

Alcohol effects estimated 3-10x FAS

Alcohol Use in Pregnancy & FAS

Alcohol is a teratogen

Timing of use

Dose of alcohol

Pattern of use

Individual risk/protective factors

Who is at Highest Risk to Have Children with FASDs?

One Drink with Dinner?

Alcohol is a known teratogen. “No safe amount of alcohol”

No clear evidence that 1 drink or less per day has caused damage either, unless using averages

Europe more permissive, up to 1 drink/day

Transparency vs consistency of message,
Public vs private discussions

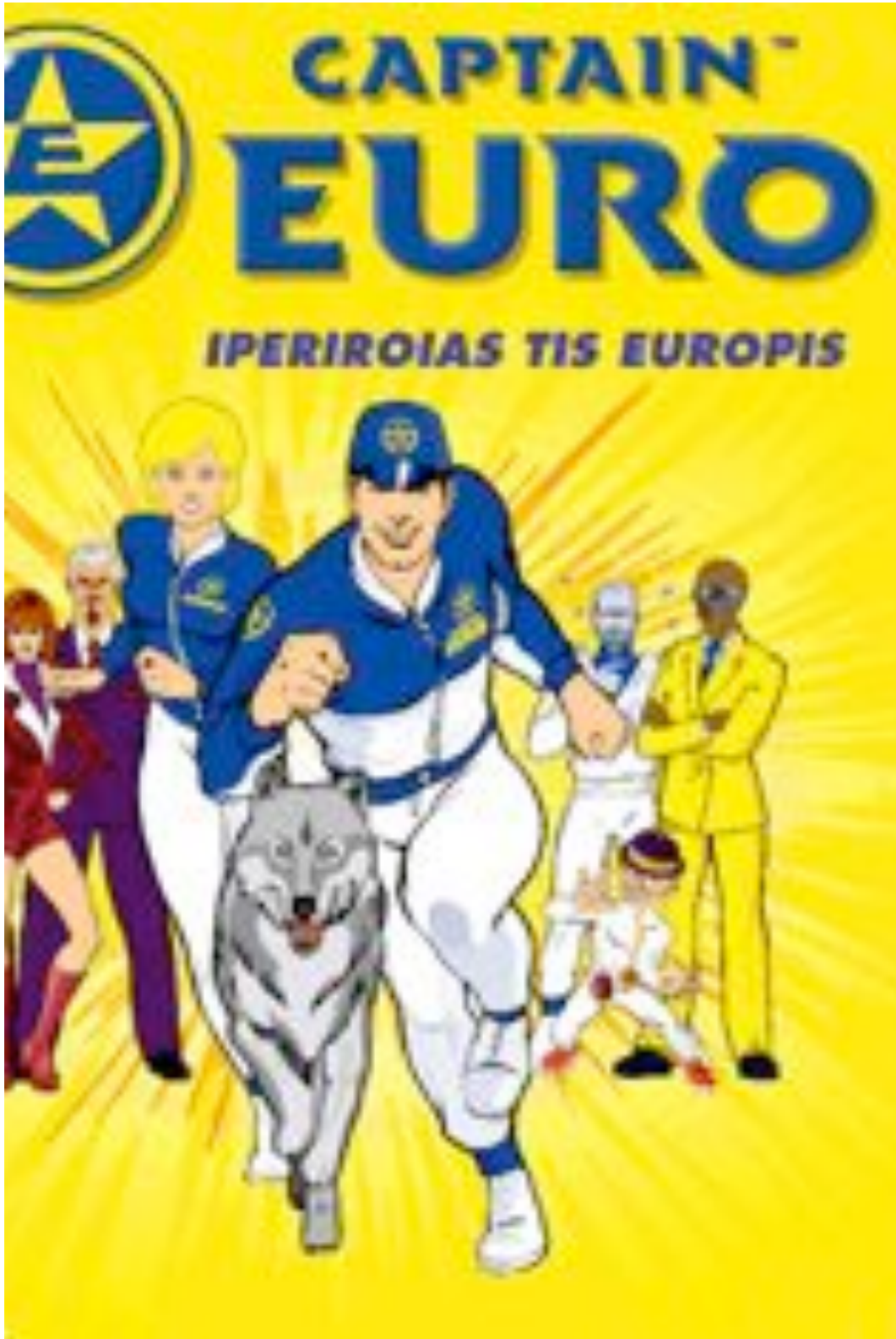
Fetal Alcohol Powers?

Kelly et al. report that “light-moderate” prenatal alcohol exposure is not assoc with behavioral/cognitive deficits at 3yo, 5yo

Actually, they do better than non-exposed kids?!?

Don’t believe the media hype (Danish study too)

Major methodologic concerns



Diagnosing FAS …

What EVERYONE Agrees On: FASD involves ...

Growth deficiency

Facial anomalies

Organic brain damage

Alcohol exposure

What Everyone DISAGREES On: The DEFINITION of ...

Growth deficiency

Facial anomalies

Organic brain damage

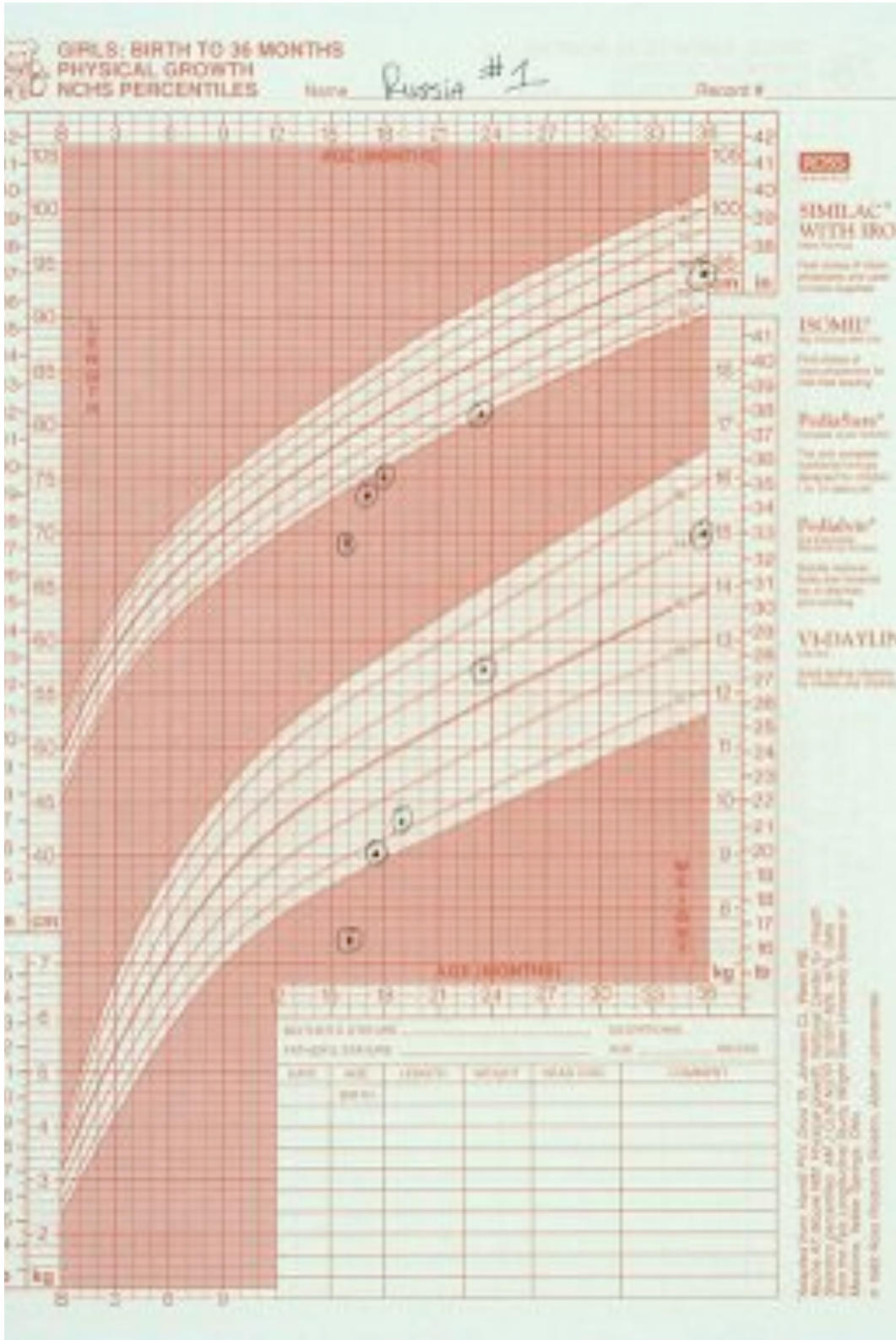
Alcohol exposure

Growth Deficiency

Height deficiency (birth or since)*

Weight deficiency (birth or since)*

*Not better explained by other influences on growth (chronic illness, severe malnutrition, etc)



Facial Anomalies

1. Small palpebral fissures

2. Smooth philtrum

3. Thin upper lip

Others are inconsistent and change with age

These probably don't

Sentinel Facial Features: Philtrum & Lip

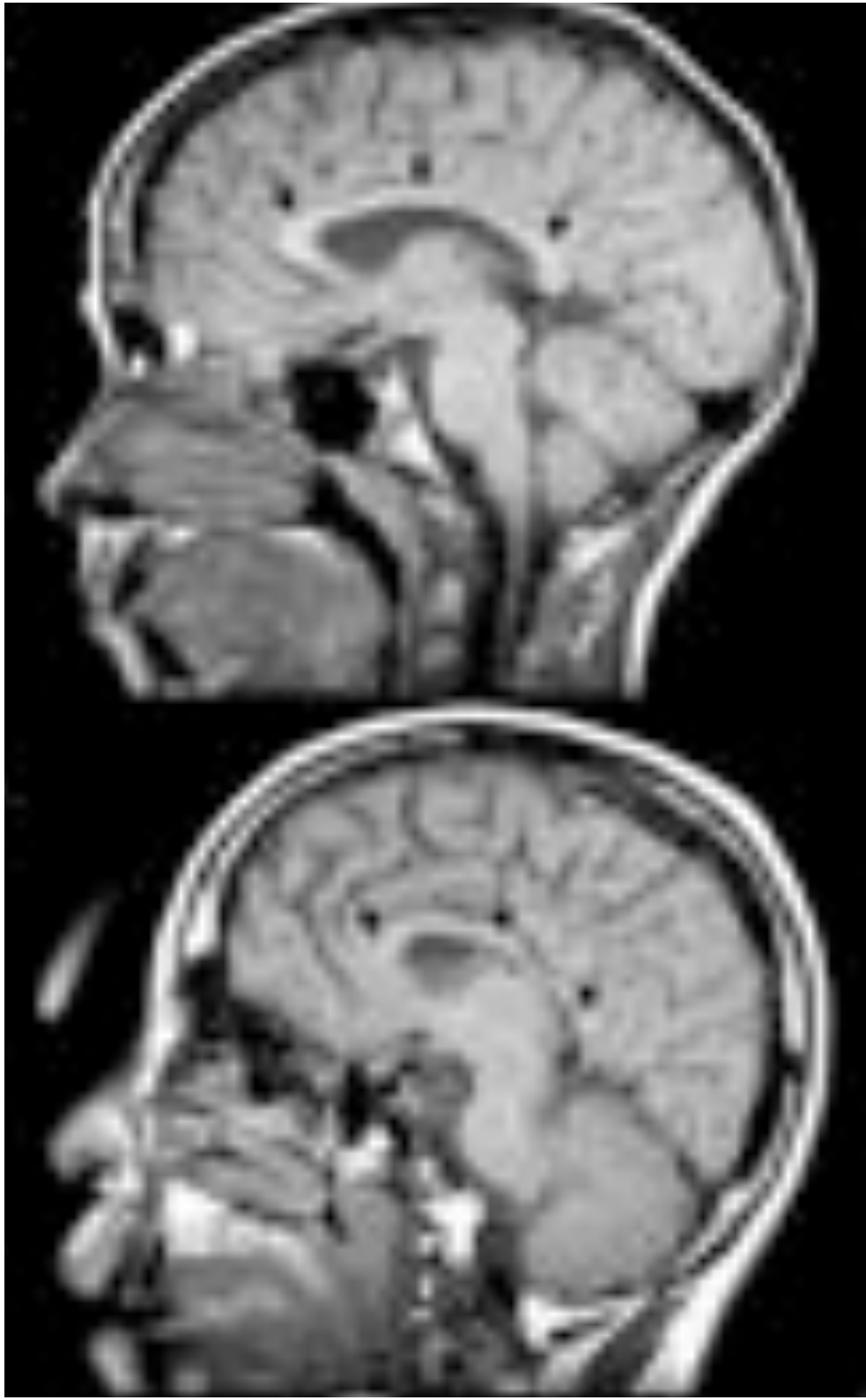
Evidence of Brain Damage

Microcephaly

“Hard” Neurologic signs, e.g. seizures

Functional Evidence of Brain Damage

- “3 strikes” criteria



Other Organ Systems

Eye: Myopia, strabismus, ptosis, optic nerve hypoplasia

ENT: Hearing impairments, clefts, micrognathia, external ear anomalies, recurrent/chronic ear infections

Cardiac: ASD/VSD, PS, PDA, AS, Tet, etc. Septal defects make up most of ARBDs in a recent study

Renal: hydronephrosis, dys/hypoplastic kidneys

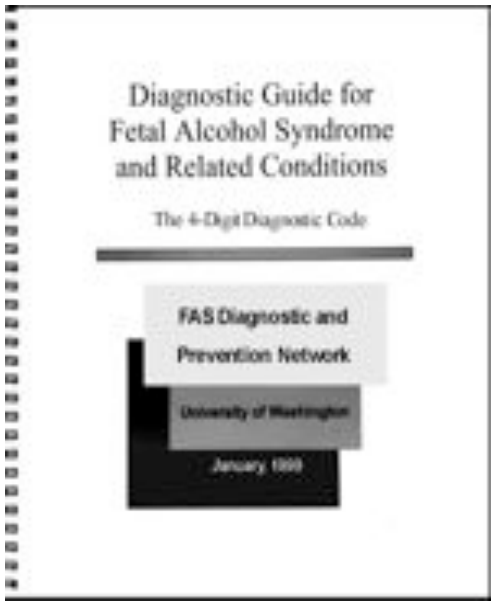
Skeletal: clinodactyly, limited ROM, pectus, scoliosis, etc

BUT - inconsistent definitions of FAS, no controlling for other risks, many not found in recent survey (overall, ARBDs 4x more likely in heavy 1st trimester PAE)

Bottom line: Hearing & vision eval, watch for heart & ENT issues

	IOM (1996)	4-Digit Code (2004)	CDC (2004)	Canadian (2005)	Hoyme (2005)
Growth	At least one. Low weight for height Low birth weight Decelerating weight	Pre- or postnatal height or weight < 10% (Growth Ranks 2, 3, or 4)	Pre- or postnatal height or weight < 10%	At least one. Pre- or postnatal height or weight < 10% Low weight-to-height ratio (= 10%)	Pre- or postnatal height or weight < 10%
Face	Characteristic pattern that includes features such as: Short PFL Flat upper lip Flattened philtrum Flat midface	All 3 required: (Face Rank 4) PFL < 3 % Philtrum (Rank 4 or 5) Lip (Rank 4 or 5)	All 3 required: PFL < 10 % Philtrum (Rank 4 or 5) Lip (Rank 4 or 5)	All 3 required: PFL < 3 % Philtrum (Rank 4 or 5) Lip (Rank 4 or 5)	2 of the 3 required: PFL < 10 % Philtrum (Rank 4 or 5) Lip (Rank 4 or 5)
CNS	At least one **bold** feature: **Structural** OFC < 3% (microcephaly) Abnormal structure and/or **Neurological** Hard/soft signs	At least one **bold** feature: **Structural** (Brain 4) OFC < 3% (microcephaly) Abnormal structure and/or **Neurological** (Brain 4) Seizure disorder Hard signs and/or **Functional** (Brain 3) 3 or more domains >2 SDs below the mean Global deficits	At least one **bold** feature: **Structural** OFC < 10% Abnormal structure and/or **Neurological** Seizure disorder Hard/soft signs and/or **Functional** 3 or more domains with impairment >1 SDs below the mean Global deficits	3 or more domains impaired: **Impairment =** Scores >2 SDs below the mean 1.5 to 2 SD discrepancy among subtests >1 SD discrepancy between subdomains	At least one **bold** feature: **Structural** OFC < 10% Abnormal structure

”4-Digit Code” Tools and Software



Lip-Philtrum Guide 1

Lip-Philtrum Guide 2

Foster Care Screening (Astley, ’02)

600 kids entering foster care in King County screened with photos, OFC

- 10 screened positive->clinic for confirmation

Nested into preexisting system (Health and Education “passport”)

Even screen-negative subjects assessed

Photo analysis was amazingly accurate – 100% sensitive, 99.8% specific

Children and families benefited

Diagnosis may get more accurate

Neurology guidelines for global dev delay recommending MRI

Neuroimaging will likely serve as a more sophisticated “head circumference”

It may also suggest specific testing and interventions

New standards are being published for normal pediatric MRI/S values

What these new tools can and can’t do

Currently useful for comparing GROUP differences in brain structures, chemistry, and function

Imaging alone cannot diagnose FASD

Overlaps between “normal” and “abnormal,” and with other conditions

Normal pediatric values not well established (but will be ...)

Bottom line: not for routine clinical use ...
Yet.

Consultation for Diagnosis

FASD team, neurodevelopmental clinic, neurology, genetics for medical aspects

Early intervention teams, psychologists, educational teams for brain function

Endocrine or growth workup for unusually severe/persistent growth problems

Neurology with imaging for microcephaly or “hard” signs, with EEG if seizures suspected

Dysmorphologist/genetics if diagnosis is uncertain

CGH Array for atypical FAS, FASD with intellectual disability?

FASD and the Brain

We hope to better understand the structural, cognitive, and behavioral features of FASDs

Brain injuries often go undiagnosed and unserved

It’s not the face that needs services

FASD is often an “invisible” disability

Children often fail to qualify for services until later school years – too late!

Variability is the rule

- For the fetal alcohol spectrum and for the child

As pediatricians and parents, we're all searching for ways to understand …

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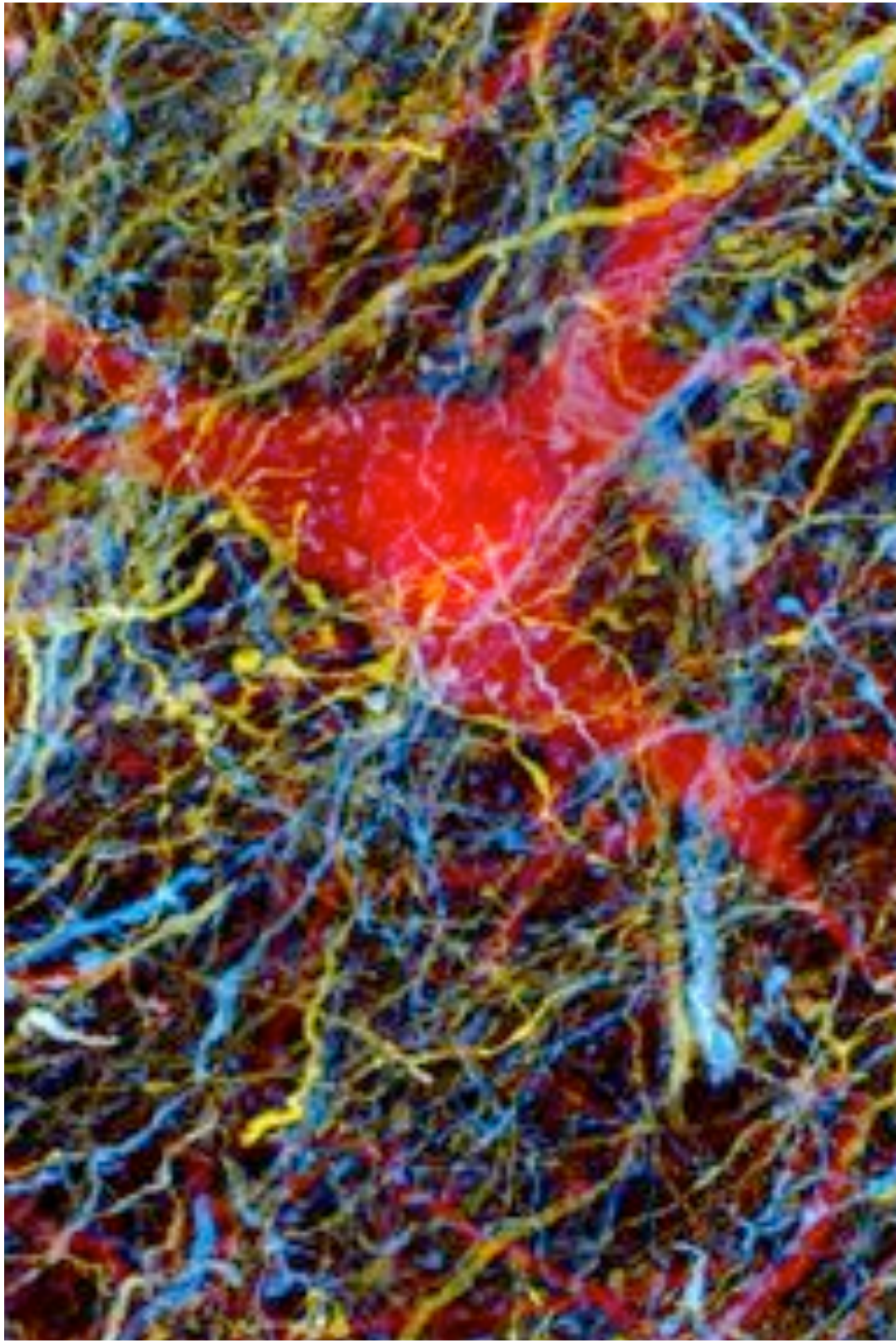
what's "normal" and what's not

what's FAS and what's something else

what's "WON'T" and what's "CAN'T"



Cognitive/Behavioral Phenotype?

Not yet (or ever?), but FASDs seem to involve:

a generalized deficit in processing complex information

(such as diminished intellectual function, slow processing, relative difficulty with complex tasks)

variability

(for the child and the spectrum)

adaptive gaps that widen with age

(can the gaps close with intervention?)

Stress, Neglect & Maltreatment

Before we blame it all on drink & drugs … don't forget other influences, like complex trauma

Prenatal stress and anxiety may also affect postnatal stress response, IQ, ADHD, sleep

Early childhood trauma can impact amygdala, hippocampus, corpus callosum, frontal lobes, cerebellar vermis

These are the same areas damaged by prenatal alcohol

Impacted Brain Domains in FASD

Cognition

Memory

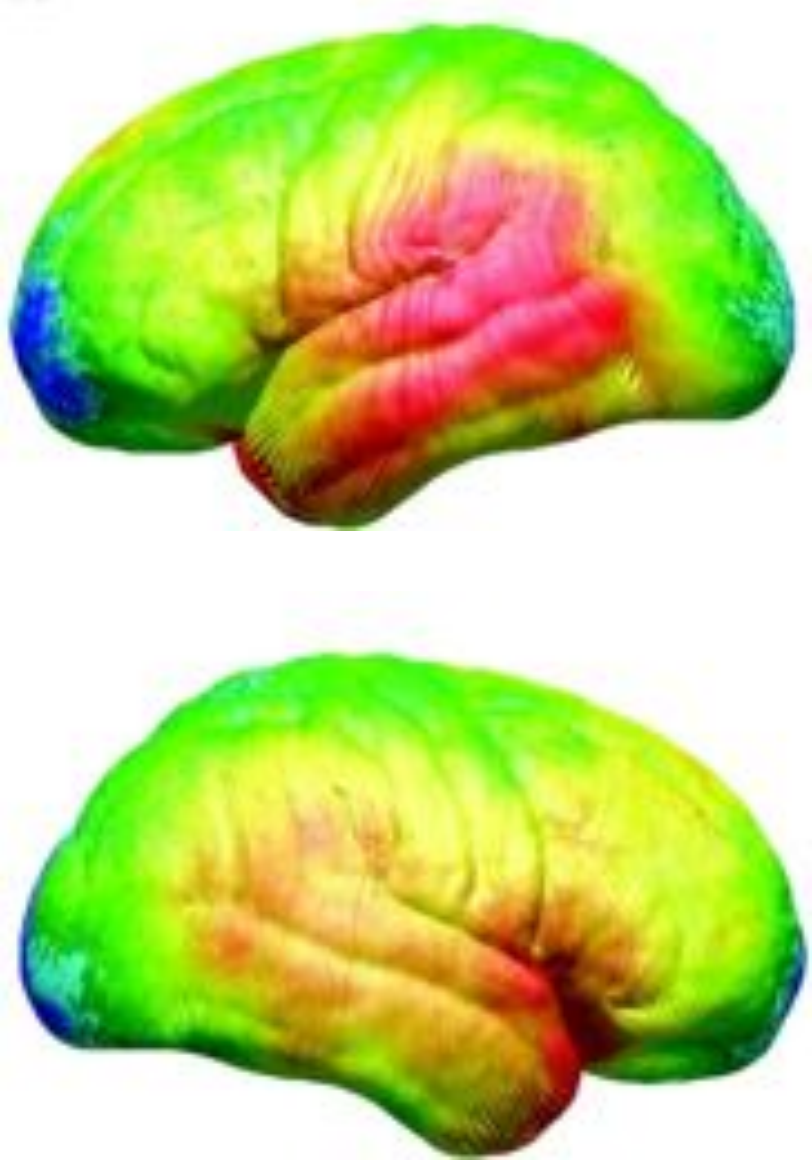
Language

Visual-motor

Executive function

ADHD

Academics



Sensory

Motor

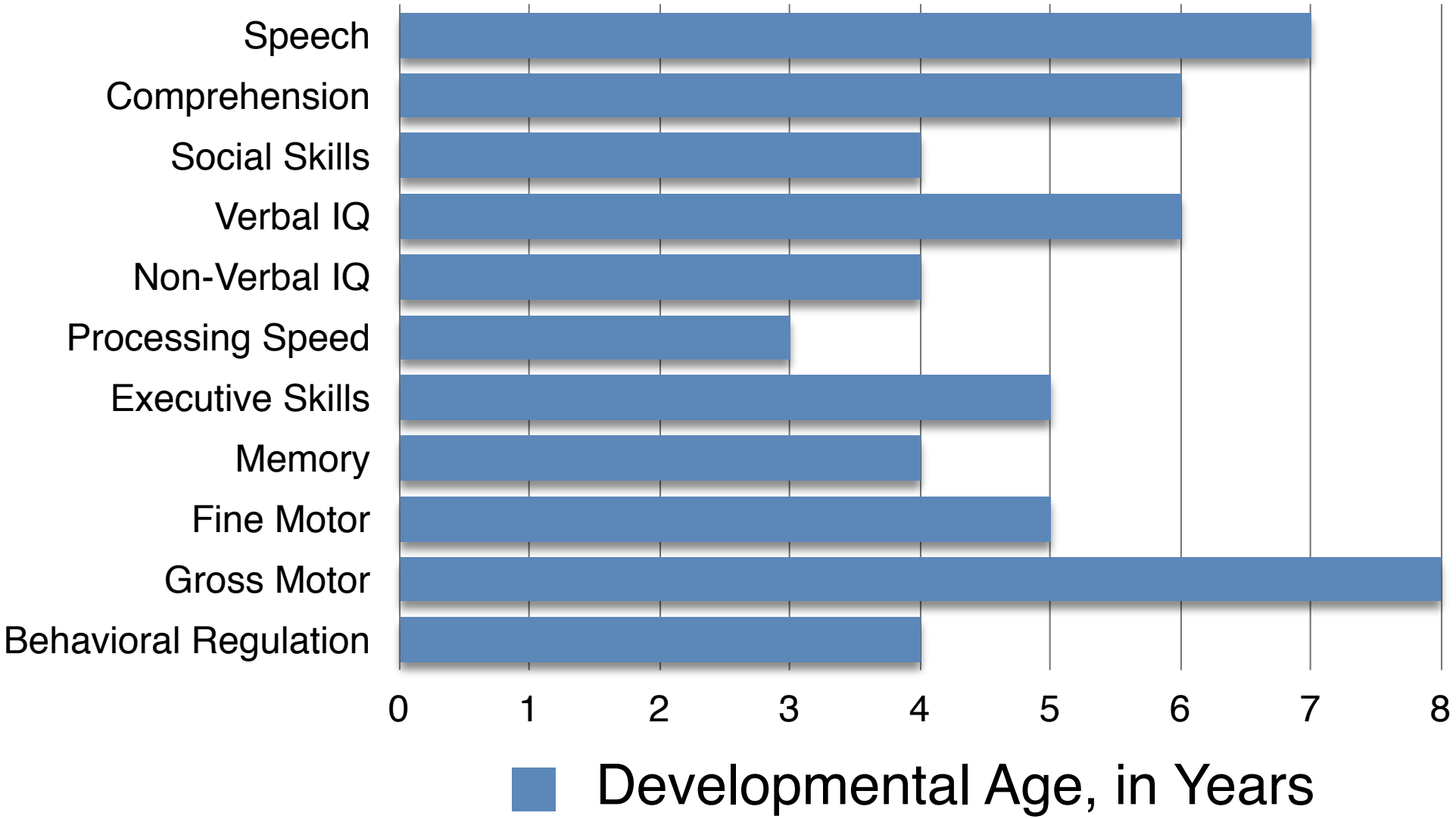
Behavior

Social Skills

Adaptive

Sleep

A Developmental Map



Newborns with FAS

Small, microcephalic?

Facial features present but trickier to assess

Often with poor state regulation, irritability, disturbed sleep, feeding difficulties, disorganized attachment

Can be a terrible “fit” – impaired parent with a very difficult infant

FASD in Preschool

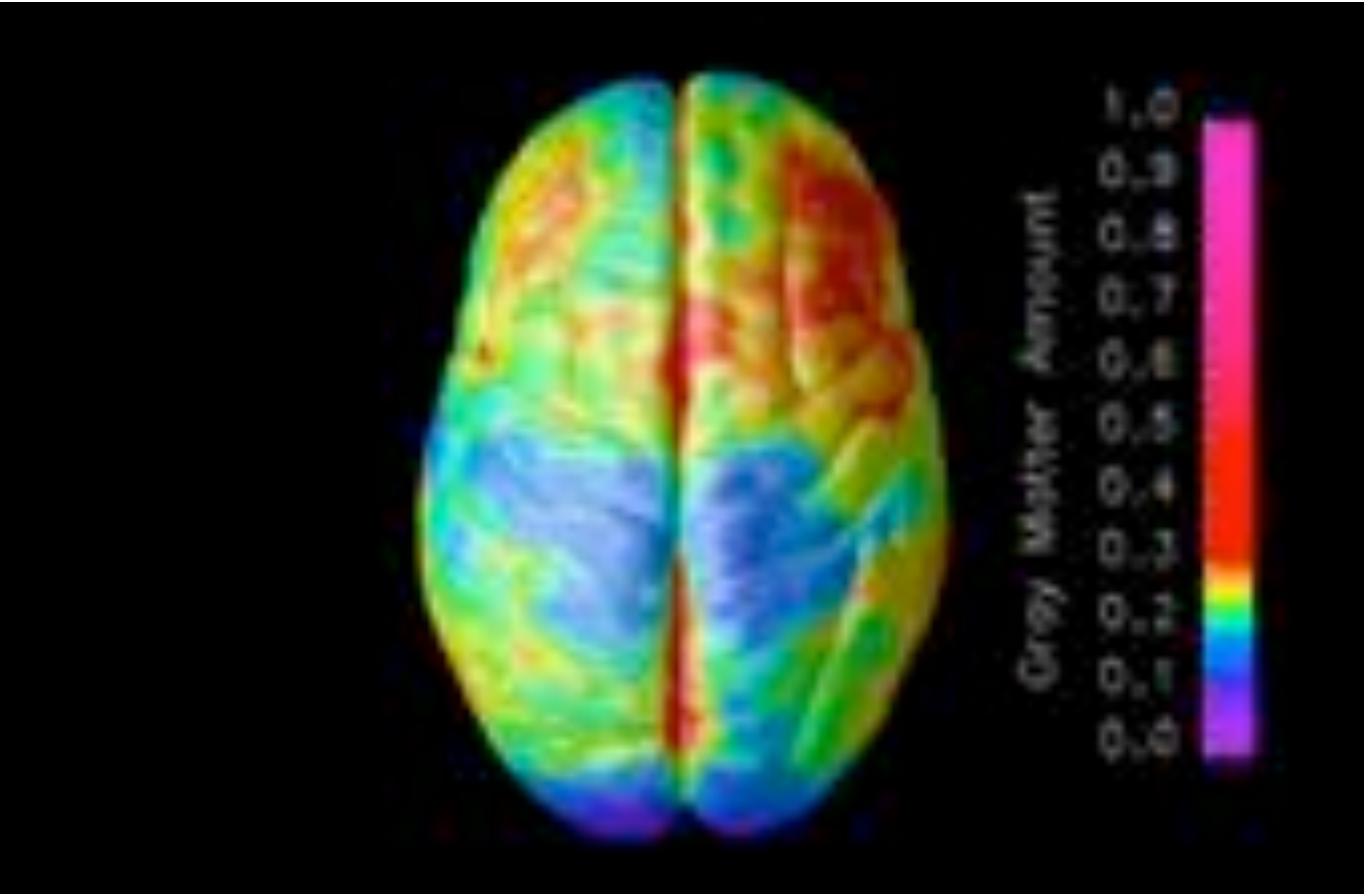
Often missed, unserved at this stage

Language, adaptive, gross and fine motor delays

Difficulty regulating emotions and behavior; extreme tantrums

Overactive, impulsive, inattentive

Slow to learn social boundaries



FASD in School and Beyond ...

Overall Cognition

STRENGTHS

Tested intelligence is usually not in retarded range

This can also be a liability, services-wise

WEAKNESSES

IQ scores lower than expected based on genetic potential

- FAS mean IQ – 66 to 80 in various studies
- FASD more variable

Lots of sub-test variability, verbal IQ vs performance IQ “splits”

Slower information processing

Nonverbal abstract reasoning

Memory

STRENGTHS

Recall of single word vocabulary and categorical labels

Recall of visual and kinesthetic patterns

Often capable of retaining verbal information (especially if words are in a rhythm or song)

WEAKNESSES

Impaired verbal learning

Auditory sequential memory

Limited working memory span

Integration and retrieval of information and concepts

Speech & Language

STRENGTHS

Superficial conversational speech – talkative and fluent

Ability to learn vocabulary and comprehend single words

WEAKNESSES

Comprehension in complicated discussions and explanations (especially out of context)

Language less complex, more superficial, more literal than peers

Comprehension scores less than expressive

"They talk better than they understand"

Understanding directions

Social communication deficits

Visual Motor Skills

STRENGTHS

Use of color

Sculptural abilities

Ability to make direct copies, especially of simple shapes

WEAKNESSES

Visual spatial organization

Making creative, complex drawings

Handwriting

Executive Functions

The brain's CEO and executive secretary

Executive function (EF) underlies many realms of adaptive behavior

Independent of intelligence

EF develops later, and continues to mature into early adulthood …

This is good and bad

MEMORY	EXECUTIVE FUNCTION	PERCENTI
		100
		95
		90
		85
		80
		75
		70
		65
		60
		55
		50
		45
		40
		35
		30
		25
X		20
		15
	X	10
		5
MEMORY	EXECUTIVE FUNCTION	PERCENTI RANK

FASD Executive Functioning Deficits

SELF-REGULATION

The ability to stay in control of emotions (“hot EF”); awareness of how others perceive you; use of self-talk strategies to monitor self and behavior

SEQUENCING OF BEHAVIOR

Knowing when and how to start an activity, keeping track of what to do next, initiating tasks

FLEXIBILITY

The ability to shift tasks smoothly, accept change, deal with transitions appropriately, absence of rigidity

Executive Functioning Deficits (continued)

RESPONSE INHIBITION

Lack of impulsivity, ability to inhibit first “knee-jerk” response to difficult situations and think before acting

PLANNING

The ability to use mental and action steps to complete tasks, to anticipate what is needed to complete tasks, related to sequencing of behavior

ORGANIZATION

The ability to keep one’s self and materials organized, in order, predictable, etc.

Attention and Behavior

Behavior regulation/mood swings

Easily overwhelmed by stimulation

Obsessive and perseverative features

Problems with visual and auditory attention

Variations on ADHD – many will receive the diagnosis, but response to meds is variable

Risk of ADHD goes up with increasing alcohol exposure (50% if Rank 4, 30% Rank 3, 15% rank 2 in several FAS clinics … Bhatara et al.)

Academic Skills

STRENGTHS

Decoding words and oral reading

Spelling skills

WEAKNESSES

Reading comprehension

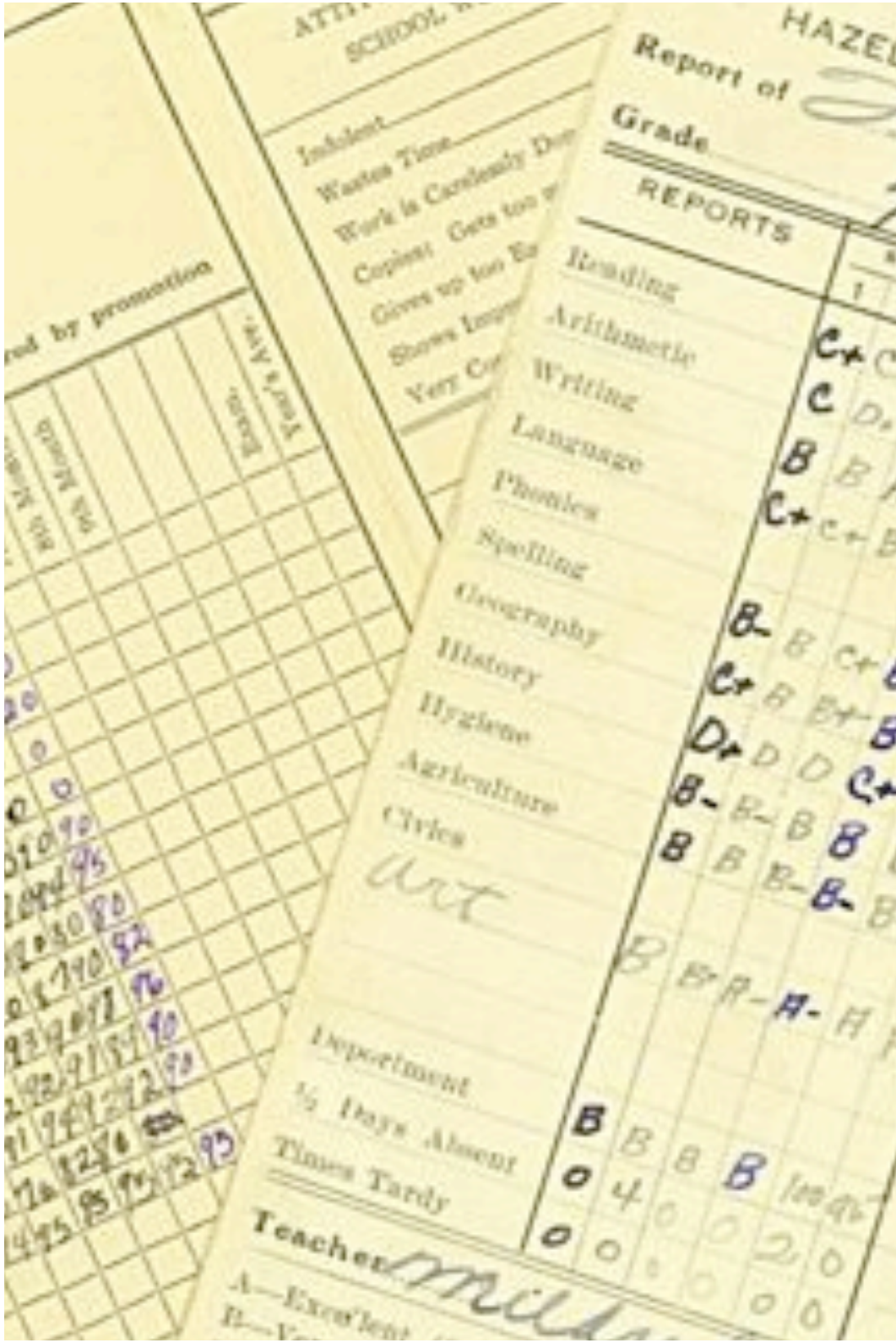
Story, essay and report writing

Arithmetic skills

Math reasoning

Organization and study skills

Academic achievement lower than IQ would predict



Sensory and Motor

Poor balance and coordination

Other “soft neurologic signs”

Visual-spatial motor skill difficulties

Sensory over-sensitivities and sensation-seeking

Athletic Skills

STRENGTHS

Individual sports requiring strength and endurance

WEAKNESSES

Team sports with demands to listen, follow directions, understand rules, and sequences and memorize procedures

Social and Behavioral Skills

STRENGTHS

Likable, friendly, engaging and often kind

Not necessarily “syndromic” in appearance

WEAKNESSES

Poor impulse control

Emotionally labile

Lack of understanding of personal boundaries

Naïve, gullible - often become a “victim”

Social/Adaptive Functioning

Social and adaptive skills often delayed, and may be half their chronologic age

This gap WIDENS in middle childhood, before it narrows

Perform better in small, highly structured environments, with range of ages

Big public high schools – OY

SAFETY! Easily victimized …

Sleep and FASDs

30-50% of our patients have problems with sleep

Rate of sleep disorders goes up with alcohol exposure

Alcohol affects circadian rhythms, the body clock

Facial anomalies and low tone make them high risk for obstructive sleep apnea

The midline cerebellum controls response to cardiorespiratory stressors

We see disturbed sleep architecture, less REM sleep

Poor sleep can mimic or worsen ADHD and other daytime cognitive and behavioral issues

Have a low threshold for sleep clinic referral

Secondary Disabilities

Are the consequences of the primary disabilities,
and arise from a gap between expectations and abilities:

Disrupted school experiences

Trouble with the law

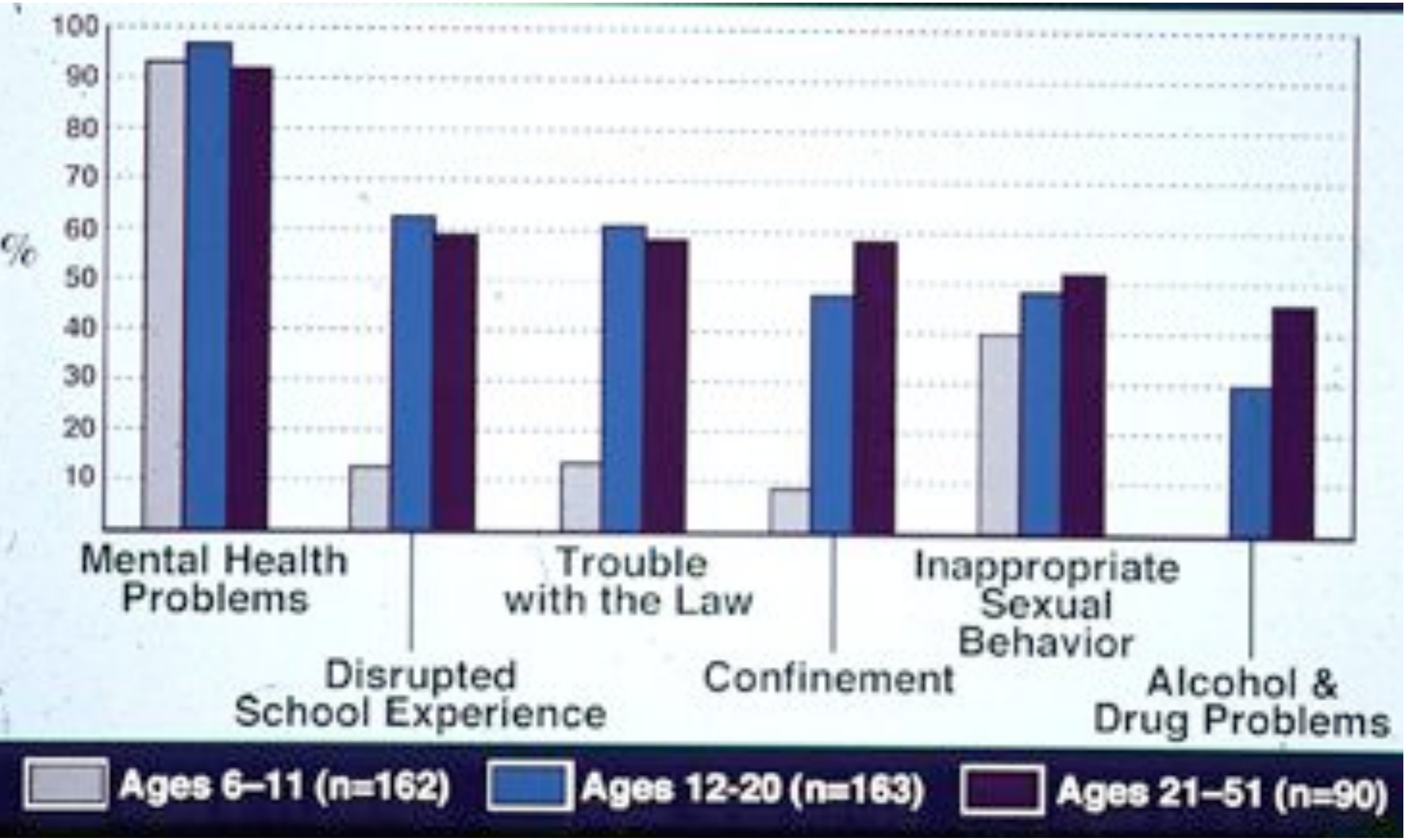
Mental health problems

Alcohol & drug abuse

Being homeless

Having children you can't care for

What we hope to prevent



Secondary Disabilities from Streissguth et al, 1996

FASD Interventions

Protective Factors Against Development of Secondary Disabilities

Early diagnosis and intervention

A caregiving environment (in middle childhood) that is:

- Nurturing, stable
- Appropriately structured & stimulating
- Geared to the child's developmental needs
- Free from caregiver substance abuse
- Safe from violence

Appropriate social services

[Adapted from Streissguth et al., 1996]

Pillars of Parenting Kids with FASDs

Structure
Supervision
Simplicity
Steps in sequence
Situational

Pillars of Parenting, Part I

Map your child's strengths and weaknesses. Start early, and repeat as they grow, as new gaps may emerge.

Be their "external brain" in areas of challenge, for as long as they need it.

Model and support self-regulation, self-calming.

Use sensory strategies to help kids maintain focus and an even keel.

Provide "scaffolding" for lagging skills.

Learning may require a lot more repetition, and since learning may not generalize to a new environment or situation, relearning may be necessary.

Pillars of Parenting, Part II

Reframe challenging behaviors as “can’t" (yet) vs “won’t”.

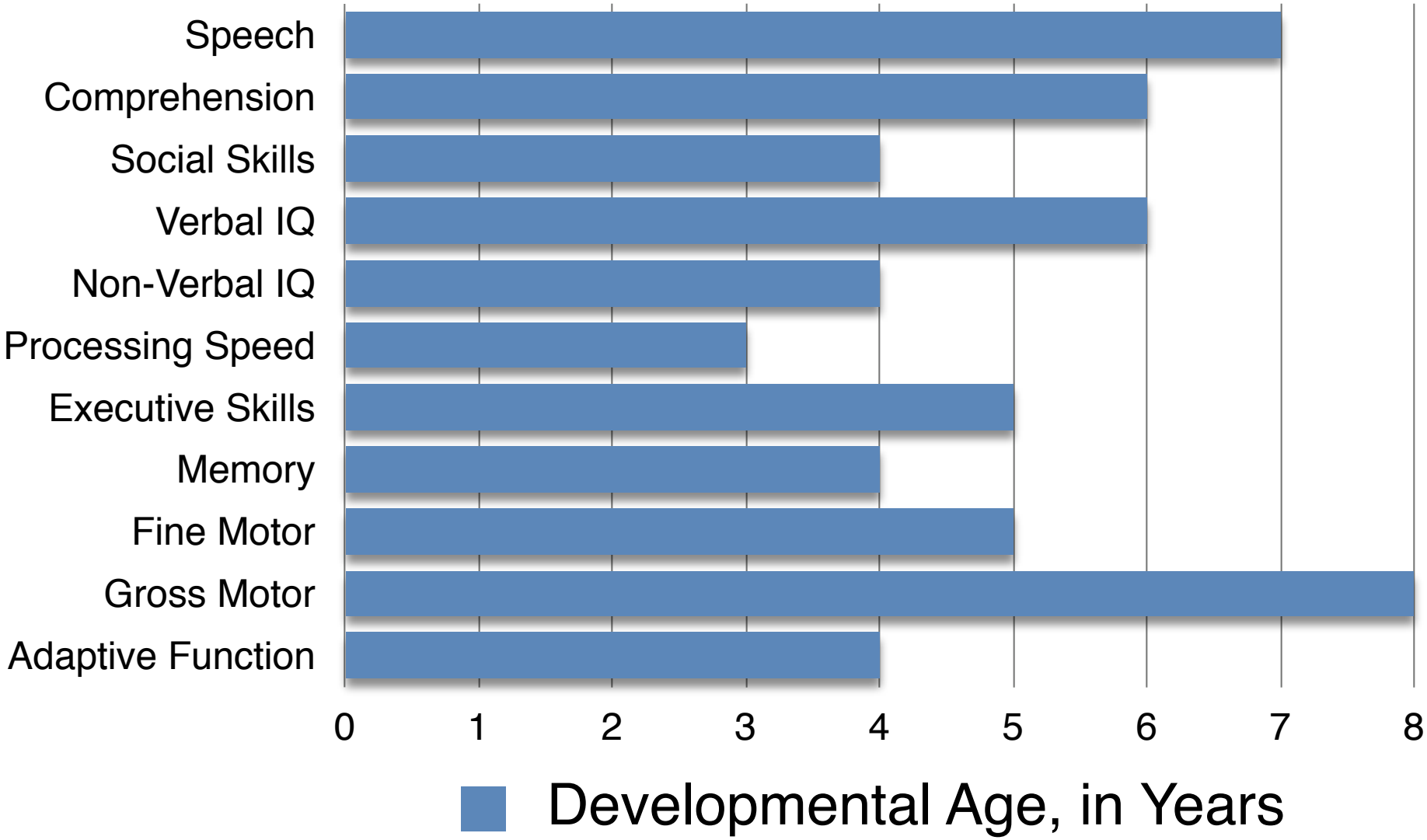
Change the environment, when you can’t change the child. Provide accommodations at home and school that reduce stress, sensory overload, help children regulate their behavior, and support their learning styles.

Use positive behavior support strategies, finding ways to prevent problem behaviors, and ways to respond that don't reinforce them.

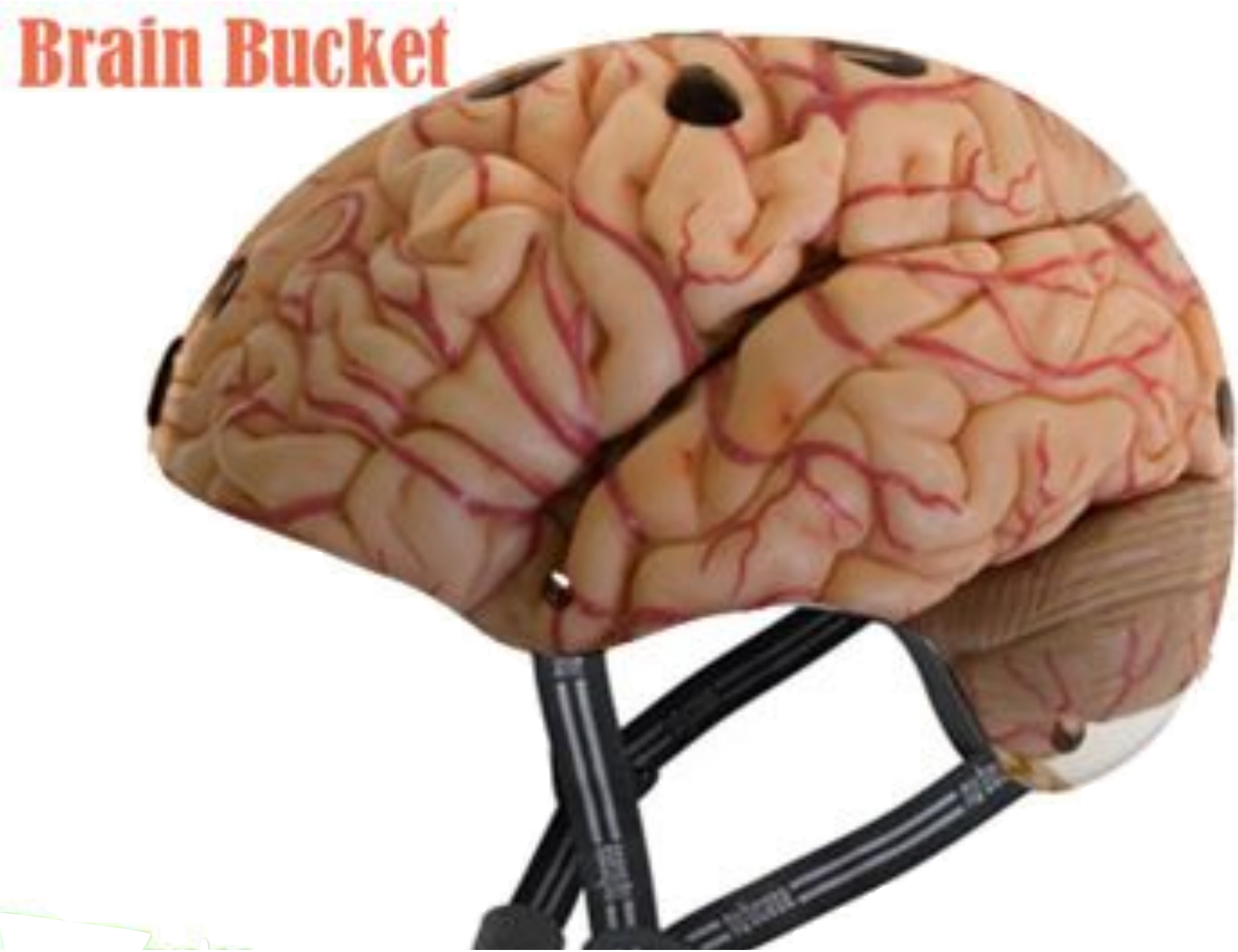
Make “invisible disabilities” visible to teachers and other caregivers.

Practice self-advocacy with your child.

Parental support and self-care is not optional.



Map, and re-map, their developmental profile



Be their “external brain” ...

Model and support self-regulation.

Daily practice, grasshopper.

Use sensory strategies

Use scaffolding for lagging skills

Learning
Relearning
Learning to learn

Reframe challenging behaviors

Change the environment and expectations

Use positive behavior support strategies ...

				Following directions the first time		*Finish task more quickly*
				Ideal Behavior	**»**	**Consequence**
Irritable after a bad day at school		*Being asked to do a chore*		*Not following directions*		*Escape task*
Setting Event/ Circumstance	**»**	**Immediate Predictor**	**»**	**Behavior**	**»**	**Consequence**
				Ask for help		*Escape task*
				Realistic Behavior	**»**	**Consequence**

Antecedents, Behaviors, Consequences (FBA, BIP)

Make “invisible disabilities” visible

Practice self-advocacy

Parent support and self-care is not optional

Intervention Research Projects

Self-Regulation & Sensory Strategies

- The Alert Program (Children's Research Triangle)

Learning How to Learn - "Cognitive Habilitation"

- Math Interactive Learning Program (Marcus Institute)

Social Skills Interventions

- Children's Friendship Training (UCLA)

Behavioral Support

- Families Moving Forward (UW research)

PCIT vs Parent Support and Management

- University of Oklahoma

Consultation for Treatment

FASD team or neurodevelopmental clinic for “developmental home”?

PT, OT, and SLPs are frequently involved in assessment and treatment

School-based or private psychologists and behavioral specialists/therapists can be invaluable

Psychiatrists for med management

Social skills groups, Special Olympics, etc

Online and local support groups for caregivers

Medications?

Stimulants
Alpha-agonists
SSRIs
Mood stabilizers
Atypical anti-psychotics

BIG Cautions

Advocating for School Needs

Birth-to-3, then Child Find

504 Plan vs IEP

Develop an ally/advocate

IEP Meetings

- Check your own pulse
- Stack the deck in your favor
- Build a succession of YES's
- Make the bureaucracy work for you

If things still aren't going well ...

NICHCY.org

Treatments of the Future?

Choline - folic acid for FAS?

- In rats, prenatal and early postnatal choline improves learning, protects against future insults
- Also protects against prenatal alcohol's effects on learning and memory (but not motor), even after alcohol exposure
- May play a role in neural tube defects (spina bifida)
- Less likely to work (as well or at all?) later in childhood
- Not ready for prime time (wait for UMN study)

Aniracetam - are the Russians right?

- Postnatal treatment may reduce anxiety and reverse alcohol's memory and cognitive impacts in rats

Key Points for Caregivers and Professionals

FASDs are too often an “invisible disability”

Refer alcohol-exposed kids for early evaluation

Thorough testing is so important

Expect deficits in complexity, integration

Individualized, longterm interventions

- Reframe behaviors, adjust expectations and child’s environment
- Behavioral consultation, self-regulation, social, learning to learn
- Targeted medication evaluations
- Anticipate adolescent and adult transitions

Caregiver education, support groups, linkage, school advocacy, respite

FASD Resources

UW Publications, Diagnostic Tools, Guides and Training Programs:

- www.fasdpn.org (including an online course in 4-Digit Code)
- www.adoptmed.org/fas

Other Online Resources

- www.cdc.gov/fasd/
- http://fasdcenter.samhsa.gov/
- www.nofas.org/ (with national resource directory)

Teaching Students with FASD

- www.education.gov.ab.ca/k_12/specialneeds/fasd.asp

FAS – A Guide for Living: Parenting Children with FASD

Native American FASD Resources

Online Resources

- http://fasdcenter.samhsa.gov/nativeinitiative/resources.aspx
- http://www.ihs.gov/headstart/documents/FetalAlcoholSpectrumDisordersAmongNativeAmericans.pdf
- http://www.comingoftheblessing.com (prevention booklet)

Journey Through the Healing Circle Series

- http://www.dshs.wa.gov/ca/fosterparents/journey.asp

Gifts from the Sacred Circle (parenting curriculum)

Thank You!

Susan Astley, PhD

Julia Bledsoe, MD

Julie Gelo

Heather Olson, PhD

Allison Brooks, PhD

The FAS Clinic Team

Our Clinic and Study Families

Maria's Children (artwork)