

What is FASD?

*Omar A. Rahman, MD
Friedland Family Distinguished Professor
Director of Genetic Medicine
Professor of Pediatrics
University of Nebraska Medical Center*



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History of FAS

1700s

- Several physician groups commented on the children of alcoholics
 - “weak, feeble, and distempered”
 - “born weak and silly... shriveled and old”
- Daniel Defoe wrote “in less than an Age, we may expect a fine Spindle-shank’d Generation”

1899

- Sullivan et al. commented on pattern of birth defects with increasing severity
 - Also noted that healthier children were born to these mothers when they were imprisoned
 - Male alcoholism was not a factor



History of FAS

1968

- Lemoine et al. in France reported on physical and behavioral characteristics in 127 children exposed to alcohol prenatally

1973

- Jones and Smith described the pattern of findings in children born to alcoholic mothers and termed it Fetal Alcohol Syndrome



FAS Defined

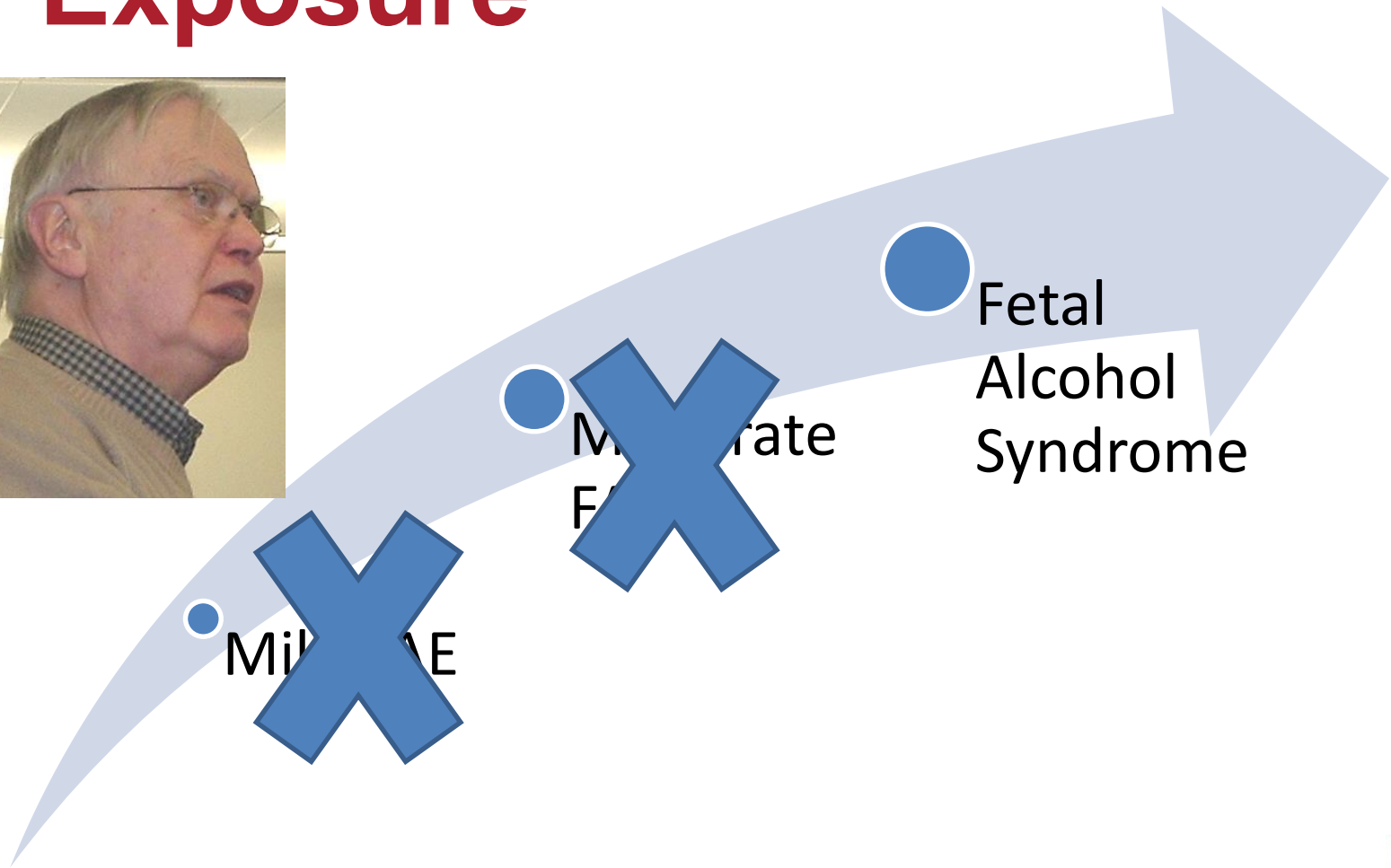
- Confirmed maternal alcohol use
- Prenatal growth deficiency
 - Length and/or weight
 - Persists after birth
- Characteristic anomalies of the face
- Neurocognitive deficits

Less severe phenotypes termed Fetal Alcohol Effects (FAE)





Spectrum of Fetal Alcohol Exposure



FASD Diagnostic Categories

From the 1996 Institute of Medicine (IOM) Report

- FAS with confirmed maternal alcohol exposure
- FAS without confirmed maternal alcohol exposure
- Partial FAS with confirmed maternal alcohol exposure
- Alcohol-related birth defects (ARBD)
- Alcohol-related neurodevelopmental disorder (ARND)

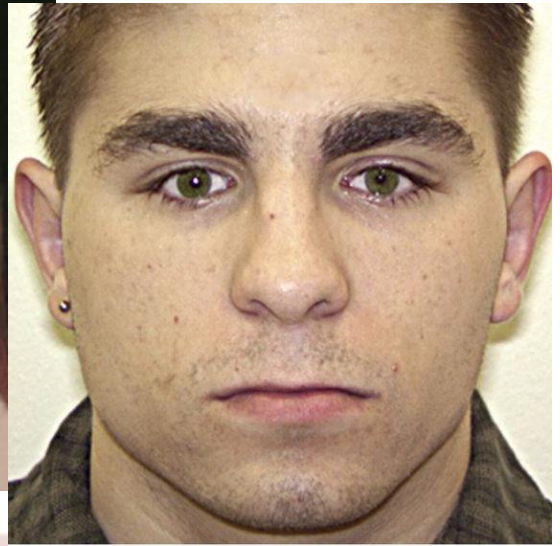
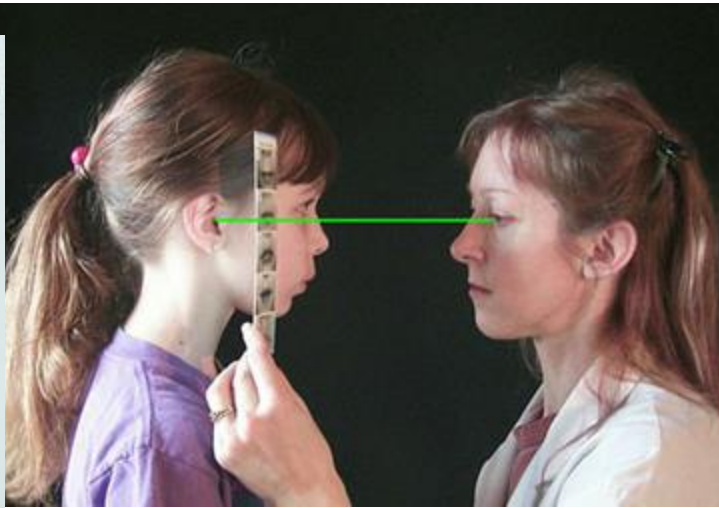
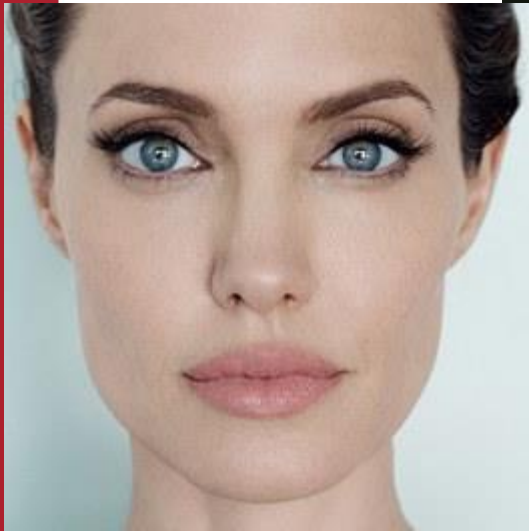


Washington Criteria

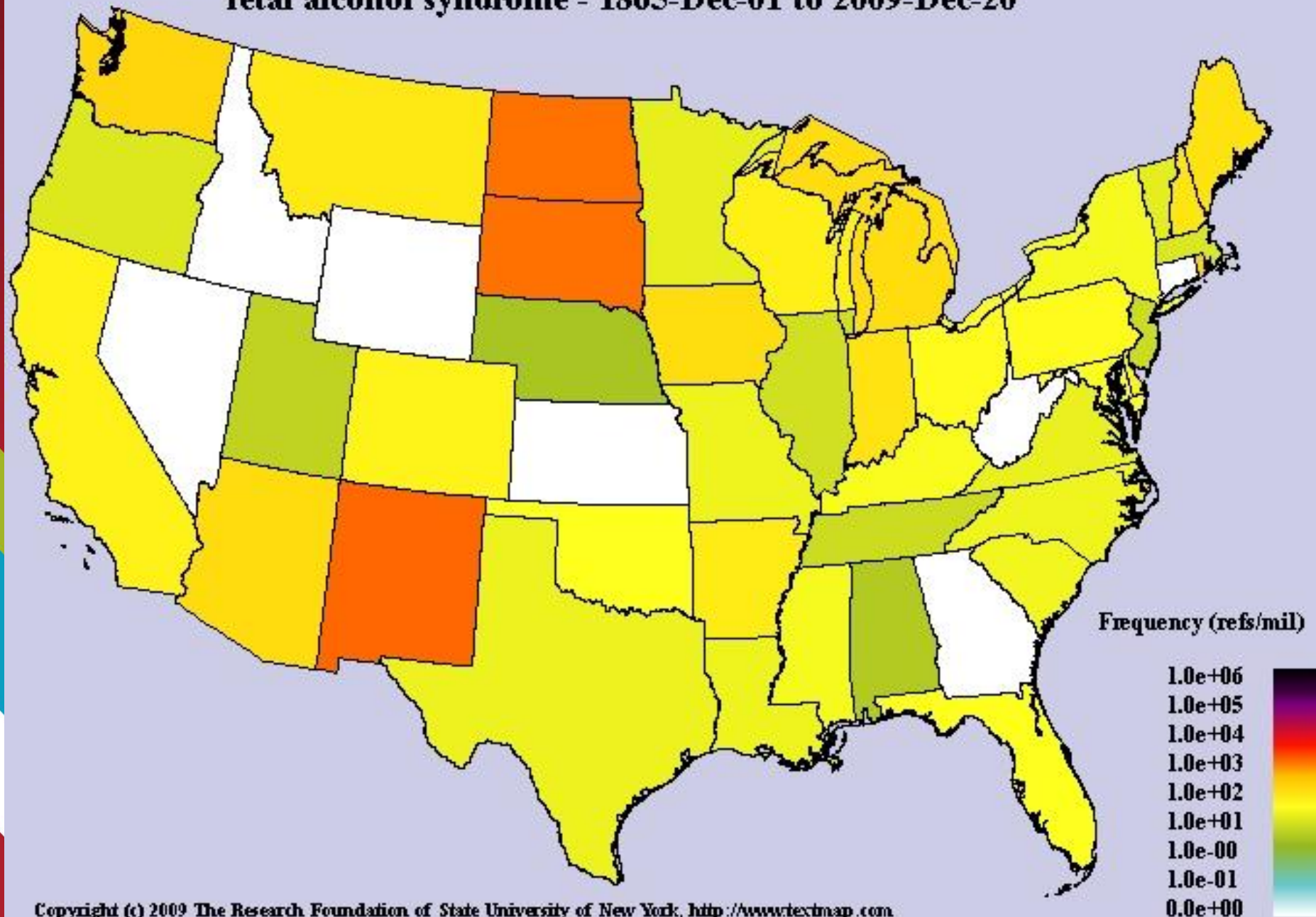
From the Clarren-Astley review of 1014 children in WA

- Use 4 variables with a score of 1-4 given for each variable
 - Prenatal growth deficiency
 - Facial features
 - Neurocognitive dysfunction
 - Alcohol exposure in utero
- 256 possible combinations from 1111 to 4444, collapsed into 22 diagnostic categories (A thru V)
- Further collapsed into 9 diagnostic outcomes by 4th variable





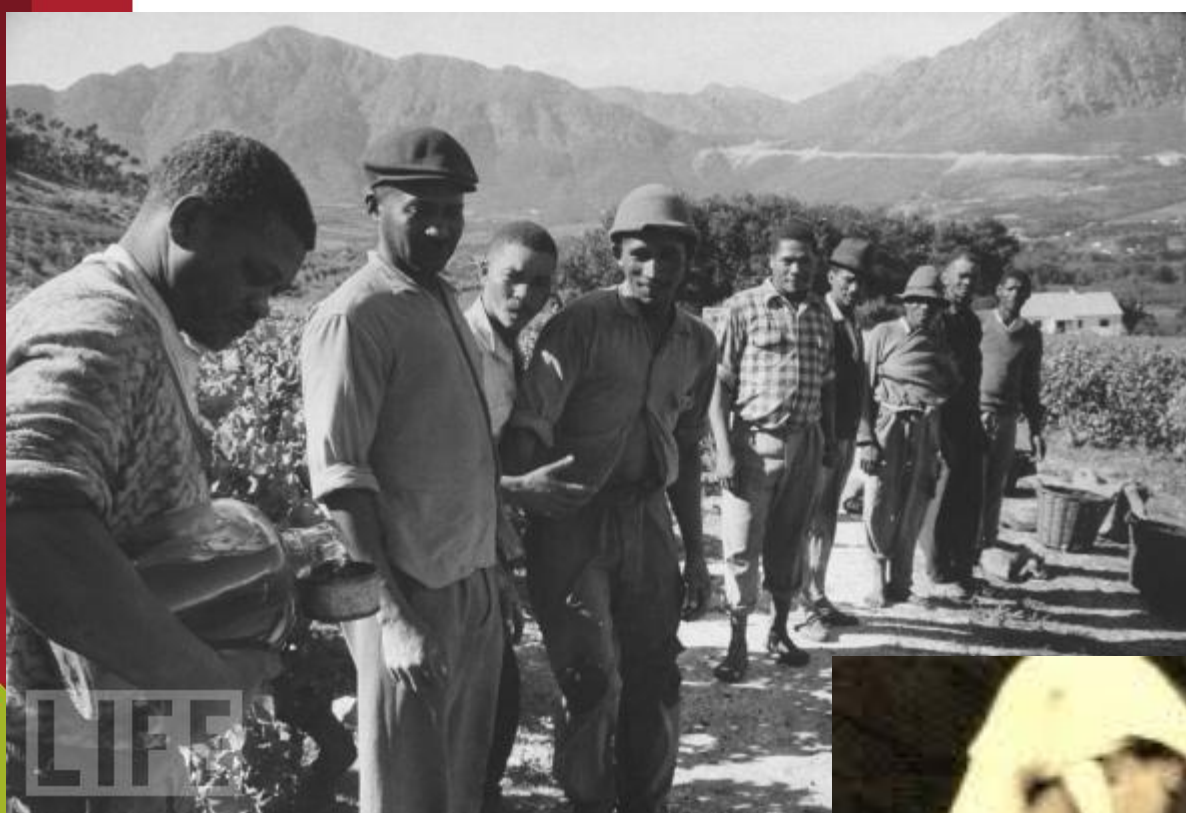
fetal alcohol syndrome - 1865-Dec-01 to 2009-Dec-20











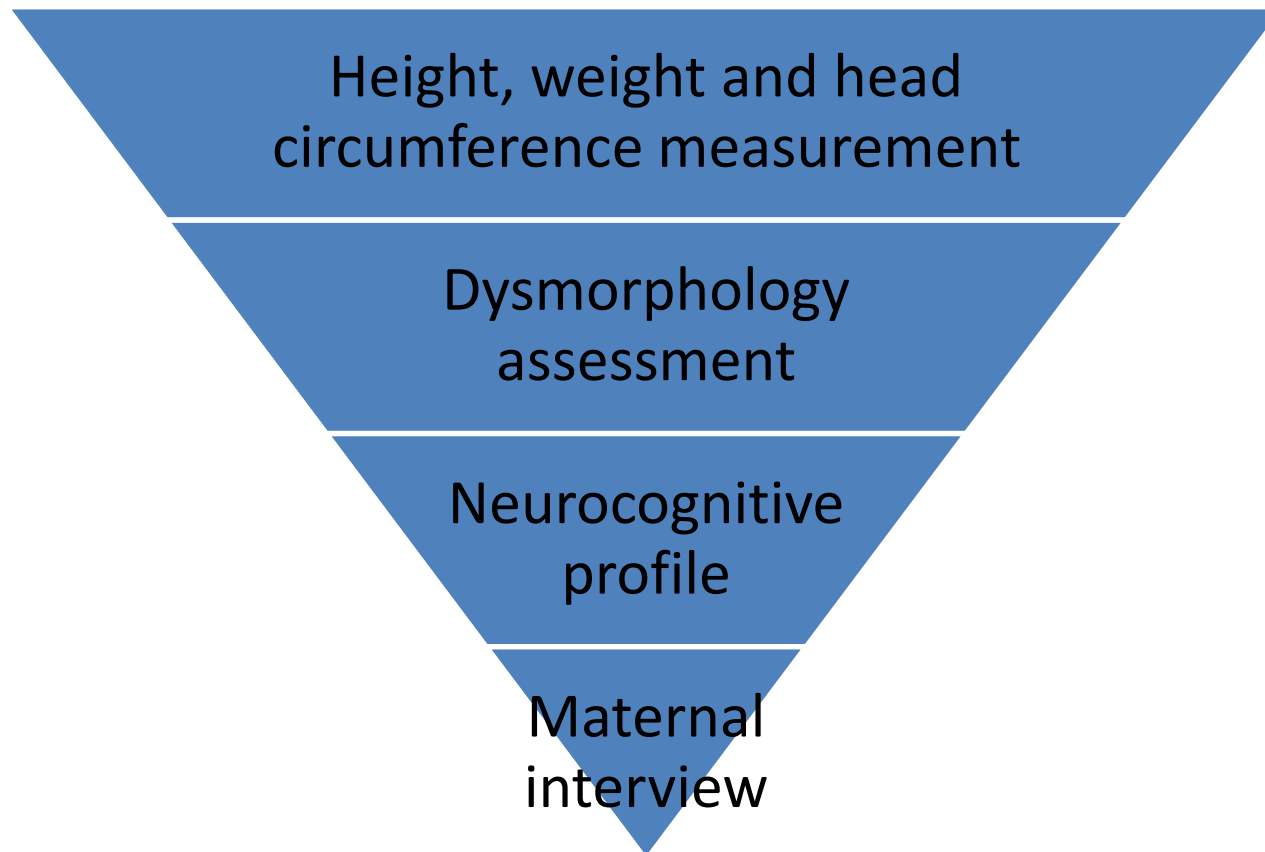


A young boy with a joyful expression is holding a white sign with a red border. He is wearing a dark blue V-neck sweater over a white collared shirt. The background shows a paved road with white dashed lines, a grassy area, and a row of palm trees under a cloudy sky. The sign contains the text: 'FAS is', '100%', and 'Preventable!!'.

FAS is
100%
Preventable!!

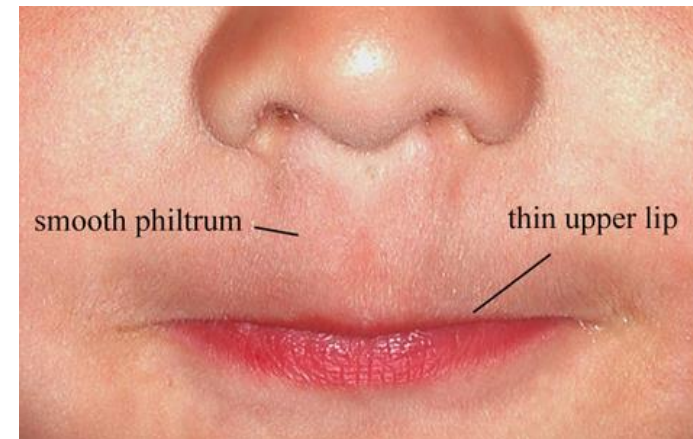
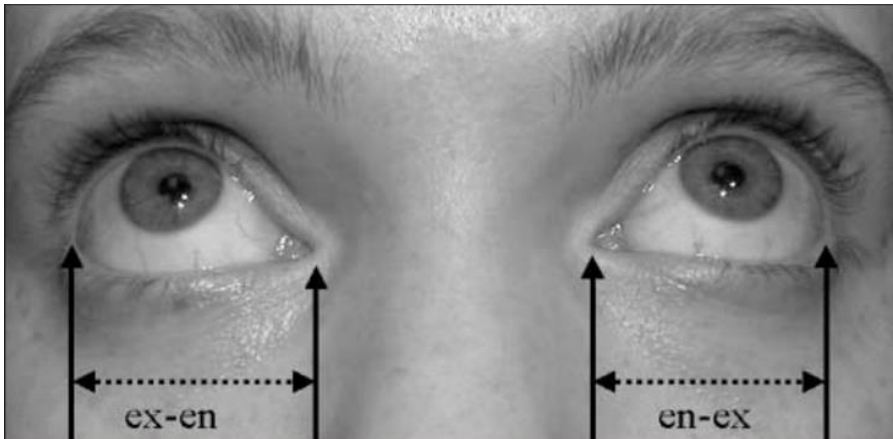
FAS School Screenings

All children in the 1st grade screened



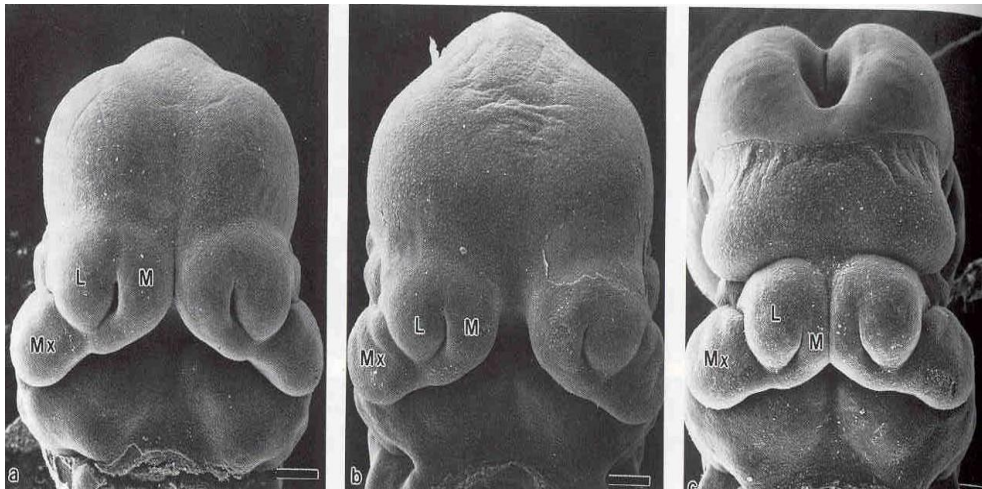
Facial assessment – cardinal findings

- Palpebral fissure length less than 10th centile
- Grade 4 or 5 lip
- Grade 4 or 5 philtrum

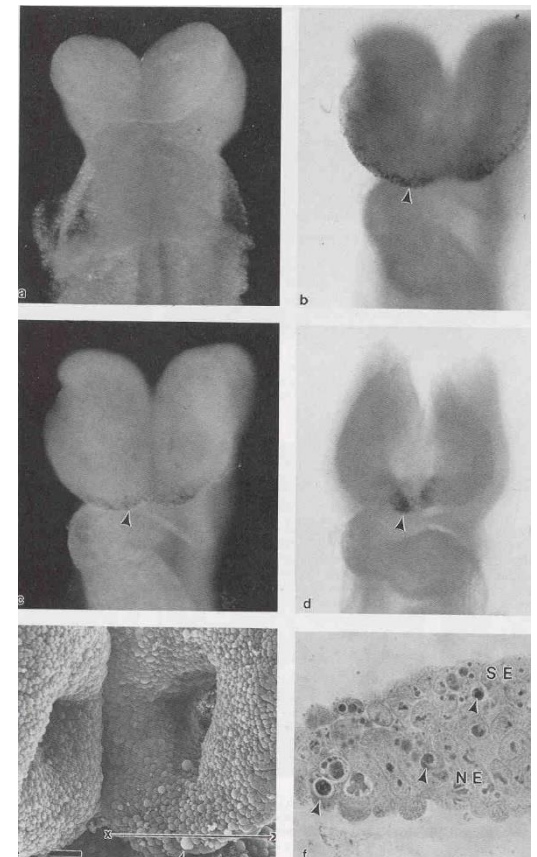


Alcohol inhibits forebrain development

ABNORMAL FRONTAL NASAL DEVELOPMENT
IN MICE
(11 DAYS)

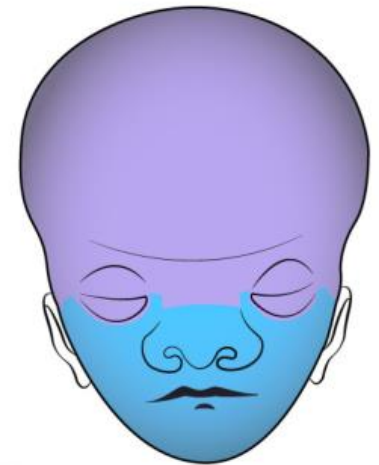
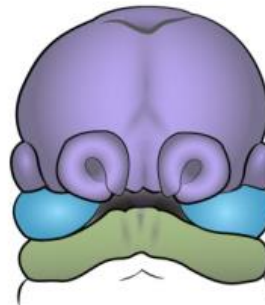
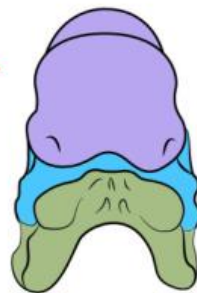


INCREASED
CELL DEATH
(8 DAYS)





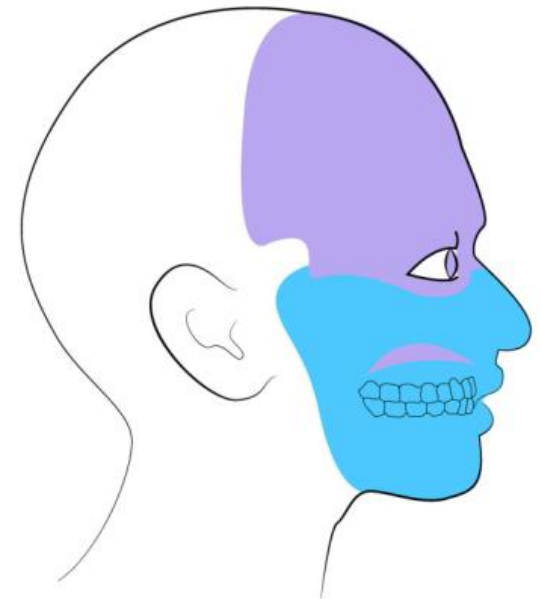
neural crest cells originate
at dorsal neural folds



neural crest cells give rise to facial ectomesenchyme
that will undergo complex morphogenesis to form a future face

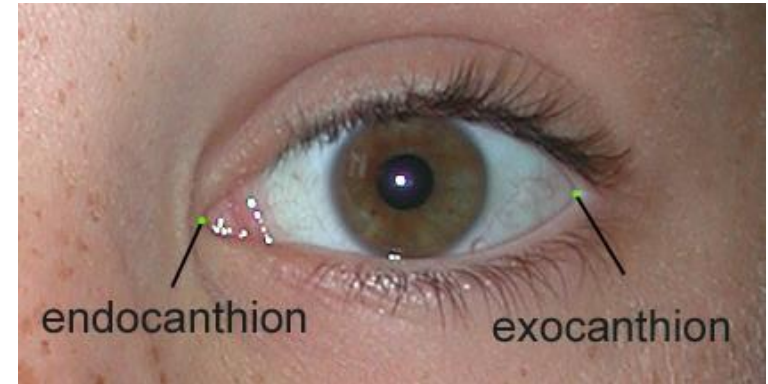


migratory neural crest populates
future facial region



Palpebral fissure length (PFL) measurement

- Document measurement from inner to outer canthus in millimeters
- Use hard, clear plastic ruler
- Follow curve of the face

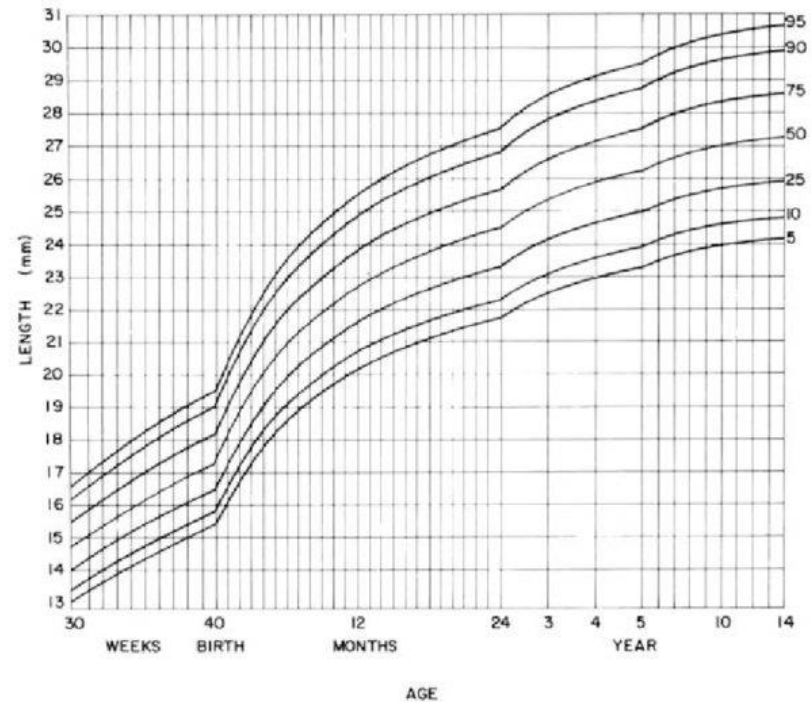


PFL

Plot measurement on graphs available in Smith's *Recognizable Pattern of Human Malformation*

Short PFL defined as <10th centile

Avoid gestalt diagnosis



Lip and Philtrum Assessment

- Ensure the patient has a neutral expression
 - Smiling will falsely increase the score
- Place guide alongside face
- Score lip and philtrum separately
- 4 or 5 is considered positive



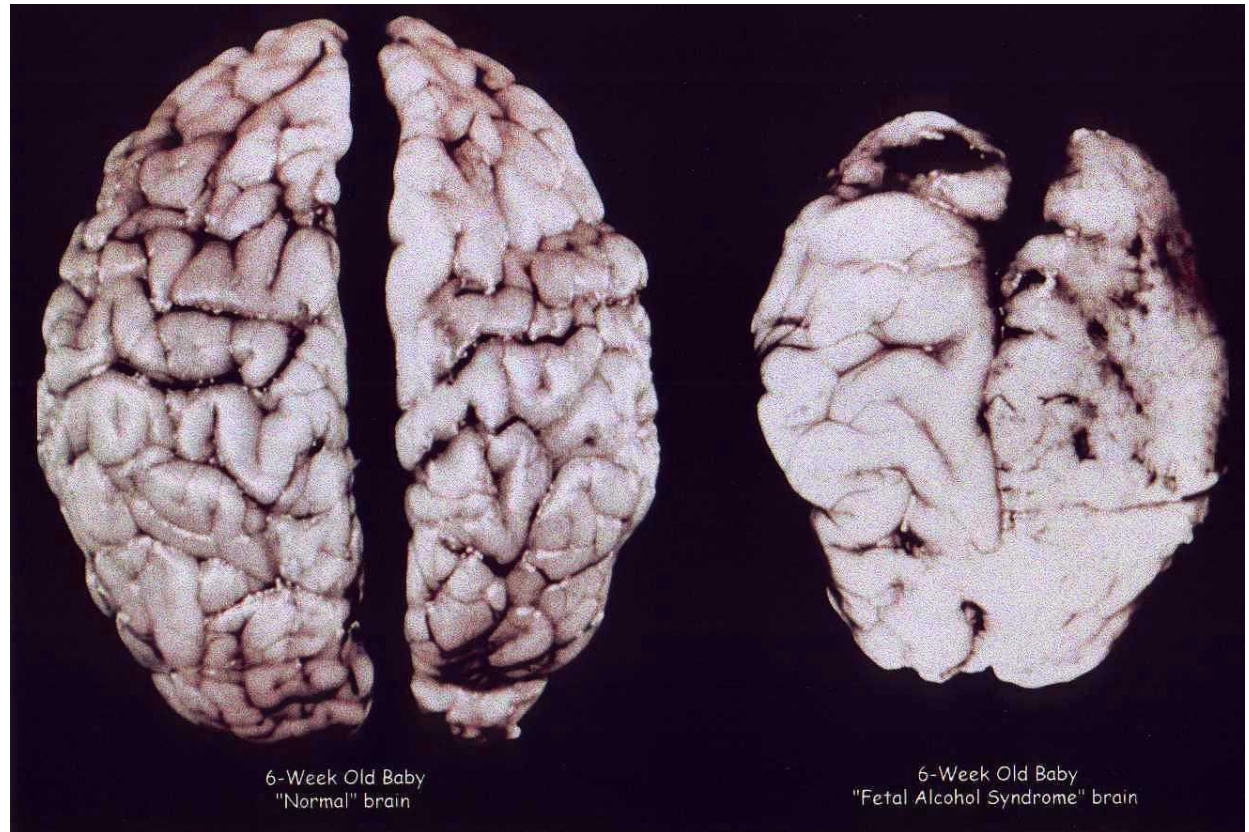


Head circumference
<10th centile

IQ testing

Behavioral and social
development
assessment

Higher executive
function assessment



Neurobehavioral assessment

Maternal interview



- Nutritional assessment conducted with over 300 items assessed
- Good nutritional status is a protective factor against the effects of alcohol
- Avoid asking “Did you drink during your pregnancy?”
 - Too accusatory
 - Uses an active verb
 - Usually elicits a negative response
- Use non-judgmental alternatives or passive verbs such as:
 - “Some women report having a beer or a glass of wine occasionally. Sometimes they have a drink before they realize they are pregnant. Do you recall anything like this happening to you?”



Maternal assessment

- Often, children are in foster care due to poor social situations
- Collateral sources of information can be used to confirm alcohol use, though may be inaccurate and based on assumption
- Try asking about alcohol-related complications such as cirrhosis, withdrawal symptoms, arrests for DUIs
- Ask about family history to identify any genetic causes of developmental delay/intellectual disability that may need to be excluded in the child
 - Intellectually disabled parents may also be predisposed to alcoholism



FASD Prevalence

Table 6. Summary of the Prevalence of FASD from In-School Studies: Rates per 1,000 Children

Diagnosis	South Africa Western Cape (1997) ^{a,b}	South Africa Western Cape (1999) ^{b,c}	South Africa Western Cape (2002) ^d	South Africa Northern Cape (2008) ^e	South Africa Mean	Italy ^f (2005)	Italy ^g (2006)	Italy Mean	USA Washington State Pilot ^h (2001)	USA Western City Pilot ⁱ (2007)	USA Western City Pilot ^j (2008)	USA Mean
FAS	(n = 42) 40.0 – 42.3	(n = 37) 40.1 – 42.9	(n = 55) 51.2 – 67.2	(n = 123) 67.2	50.1	(n = 4) 3.7 – 7.4	(n = 4) 4.4 – 9.2	6.2	(n = 7) 3.1	(n = 1) 1.4 – 2.5	(n = 5) 6.4 – 11.3	4.9
PFAS	(n = 4) 3.8 – 4.0	(n = 29) 31.4 – 33.6	(n = 18) 16.8 – 22.0	(n = 38) 20.8	18.9	(n = 17) 15.7 – 31.3	(n = 19) 21.0 – 43.8	27.9	–	(n = 6) 8.1 – 14.8	(n = 6) 7.7 – 13.5	11.0
ARND	–	(n = 10) 10.9 – 11.6	–	–	11.3	(n = 1) 0.9 – 1.8	–	1.4	–	–	–	–
ARBD	–	–	–	–	–	–	(n = 1) 1.1 – 2.3	1.7	–	–	–	–
Total FASD	(n = 46) 43.8 – 46.4	(n = 76) 82.4 – 88.16	(n = 73) 68.0 – 89.2	(n = 161) 88.0	72.3	(n = 22) 20.3 – 40.5	(n = 24) 26.6 – 55.4	35.7	–	(n = 7) 9.5 – 17.4	(n = 11) 14.1 – 24.8	16.5



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NEWS RELEASES

Tuesday, February 6, 2018

Study of first-graders shows fetal alcohol spectrum disorders prevalent in U.S. communities

NIH-funded research examined over 6,000 children to determine prevalence of FASD ranged from 1.1 to 5 percent.



A Practical Clinical Approach to Diagnosis of Fetal Alcohol Spectrum Disorders: Clarification of the 1996 Institute of Medicine Criteria

H. Eugene Hoyme, MD^{*}; Philip A. May, PhD[‡]; Wendy O. Kalberg, MA, CED[‡];
Piyadasa Kodituwakku, PhD[‡]; J. Phillip Gossage, PhD[‡]; Phyllis M. Trujillo, BS[‡]; David G. Buckley, MA[‡];
Joseph H. Miller, MSW[‡]; Alfredo S. Aragon, PhD[‡]; Nathaniel Khaole, MD[§]; Denis L. Viljoen, MD^{§||};
Kenneth Lyons Jones, MD[¶]; and Luther K. Robinson, MD[#]

Fetal alcohol spectrum disorder: a guideline for diagnosis across the lifespan

Jocelynn L. Cook PhD, Courtney R. Green PhD, Christine M. Lilley PhD, Sally M. Anderson PhD, Mary Ellen Baldwin, Albert E. Chudley MD, Julianne L. Conry PhD, Nicole LeBlanc MD, Christine A. Looch MD, Jan Lutke, Bernadene F. Mallon MSW, Audrey A. McFarlane MBA, Valerie K. Temple PhD, Ted Rosales MD; for the Canada Fetal Alcohol Spectrum Disorder Research Network

Updated Clinical Guidelines for Diagnosing Fetal Alcohol Spectrum Disorders

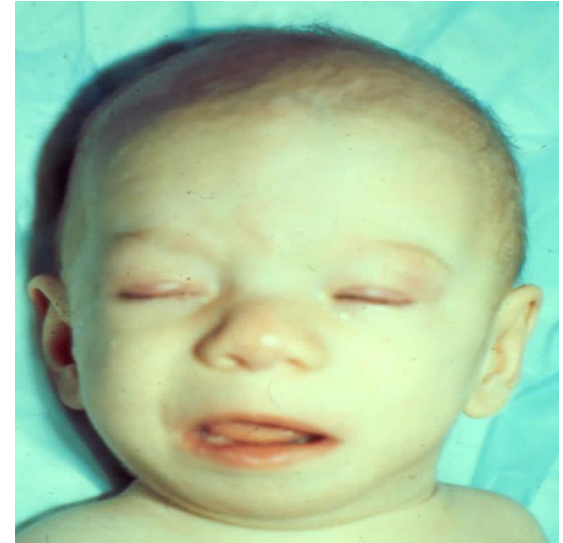
H. Eugene Hoyme, MD,^{a,b} Wendy O. Kalberg, MA, LED,^c Amy J. Elliott, PhD,^a Jason Blankenship, PhD,^{c,†} David Buckley, MA,^c Anna-Susan Marais, B Cur Nursing,^d Melanie A. Manning, MD,^e Luther K. Robinson, MD,^f Margaret P. Adam, MD,^g Omar Abdul-Rahman, MD,^h Tamison Jewett, MD,ⁱ Claire D. Coles, PhD,^j Christina Chambers, PhD, MPH,^k Kenneth L. Jones, MD,^k Colleen M. Adnams, MBChB,^l Prachi E. Shah, MD,^m Edward P. Riley, PhD,ⁿ Michael E. Charness, MD,^o Kenneth R. Warren, PhD,^p Philip A. May, PhD^{a,c,q}



Updated Hoyme Criteria

I. FAS (with or without documented prenatal alcohol exposure) requires all features A–D

- A. A characteristic pattern of minor facial anomalies, including 2/3 of the cardinal facial features
- B. Prenatal and/or postnatal growth deficiency
- C. Deficient brain growth or abnormal morphogenesis, or abnormal neurophysiology
- D. Neurobehavioral impairment
 - For children over 3 years, either one of the following:
 - With cognitive impairment
 - With behavioral impairment without cognitive impairment
 - For children less than 3 years
 - Evidence of developmental delay



Updated Hoyme Criteria

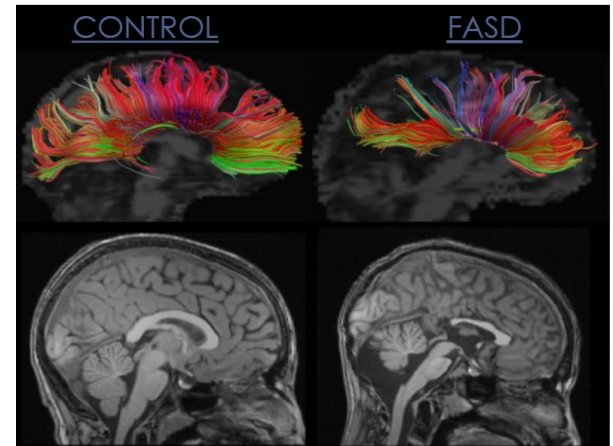
II. Partial FAS

- With documented prenatal alcohol exposure, need A and B
 - A. A characteristic pattern of minor facial anomalies, including 2/3 of the cardinal facial features
 - B. Neurobehavioral impairment
 - For children over 3 years, either one of the following:
 - » With cognitive impairment
 - » With behavioral impairment without cognitive impairment
 - For children less than 3 years
 - » Evidence of developmental delay
- Without documented prenatal alcohol exposure, need A and B from above in addition to the following
 - Growth deficiency or deficient brain growth, abnormal morphogenesis, or abnormal neurophysiology



Updated Hoyme Criteria

- III. ARND (requires both A and B, cannot be diagnosed in children less than 3 years)
- A. Documented prenatal alcohol exposure
 - B. Neurobehavioral impairment
 - With cognitive impairment
 - With behavioral impairment without cognitive impairment



Updated Hoyme Criteria

IV. ARBD (requires features A and B)

- A. Documented prenatal alcohol exposure
- B. One or more specific major malformations demonstrated in animal models and human studies to be the result of prenatal alcohol exposure
 - Cardiac: atrial septal defects, aberrant great vessels, ventricular septal defects, conotruncal heart defects
 - Skeletal: radioulnar synostosis, vertebral segmentation defects, large joint contractures, scoliosis
 - Renal: aplastic/hypoplastic/dysplastic kidneys, “horseshoe” kidneys/ureteral duplications
 - Eyes: strabismus, ptosis, retinal vascular anomalies, optic nerve hypoplasia
 - Ears: conductive hearing loss, neurosensory hearing loss



FETAL ALCOHOL SPECTRUM DISORDERS (FASD) DIAGNOSTIC CRITERIA

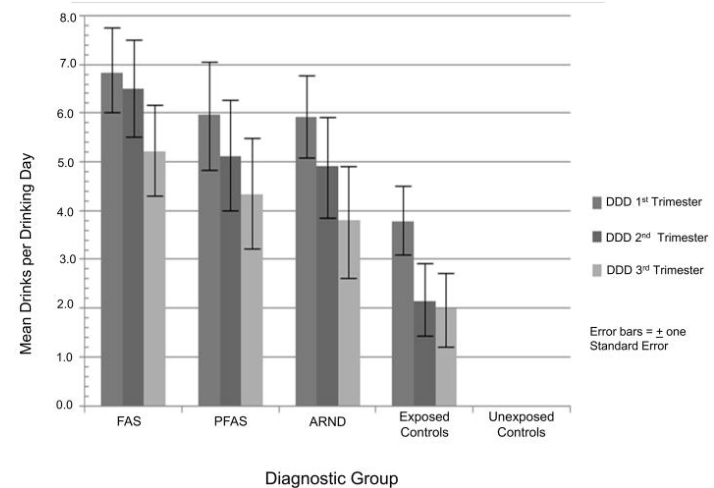
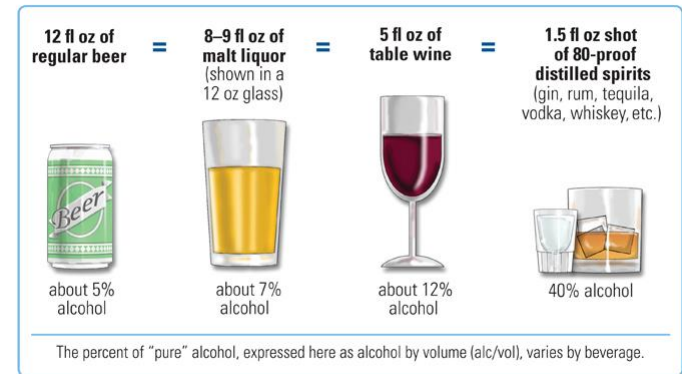
Neurobehavioral impairment	Facial features	Low height <u>and/or</u> weight	CNS anomalies	Major malformation	
WITH DOCUMENTED ALCOHOL EXPOSURE					
+	+	+	+	-	FAS
+	+	-	-	-	PFAS
+ *	-	-	-	-	ARND
-	-	-	-	+	ARBD
WITHOUT DOCUMENTED ALCOHOL EXPOSURE					
+	+	+	+	-	FAS
+	+	+ ← or → +		-	PFAS

* Neurobehavioral impairment in ARND cannot be adequately assessed in individuals under 3 years of age



Documented Prenatal Alcohol Exposure

- 1 drink is about 10-14g alcohol
- 6 or more drinks/wk for 2 or more weeks
- 3 or more drinks per occasion on 2 or more occasions
- Documentation of alcohol-related social or legal problems in proximity to the pregnancy
- Documentation of intoxication during pregnancy by blood, breath, or urine alcohol testing
- Positive testing with established alcohol-exposure biomarker (FAEE)
- Increased prenatal risk associated with drinking during pregnancy assessed by validated screening tool



Timing of exposure

Week 1-3

- "all-or-none": the embryo either dies or survives without any complications

Week 3-8 (Embryonic Period)

- most vulnerable time period due to organogenesis

Week 4-14

- fetal face development

Week 8-38

- growth and function of organ/embryo is affected
- decreased susceptibility due to organs already formed



14
(32 days)



17
(41 days)



23 (56 days)



We believe that we can measure the humanity of any society by how it takes care of individuals with intellectual and developmental disabilities.



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