

Identification and Diagnosis of Adolescents and Adults with FASDs; Establishing Diagnostic Centers and Building Capacity; Diagnostic Challenges

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Topics and threads

- Review FAS as a reference point
- Identify challenges and unanswered questions related to the diagnostic process, with emphasis on issues particular to the evaluation of adolescents and adults
- Summarize “system” needs

FAS Diagnostic Criteria

- Growth Retardation*
- Specific Facial Anomalies*
- Central Nervous System
Impairments (structural or functional)

*Corrected for racial norms if possible.

FASD's - Effects on growth

- Reduced birth weight (~160g for each ounce of alcohol consumed per day); weight gain *may* improve in childhood, *often* increases in adolescence and adulthood
- Mild short stature from birth; *usually* persists
- Microcephaly (small head size) at birth; *usually* persists or worsens with time, but *occasionally* resolves (counts also as a CNS effect)

Growth Impairment

- Pre- and/or postnatal growth impairment:
 - Height or weight at or below the 10th percentile *at any time.*



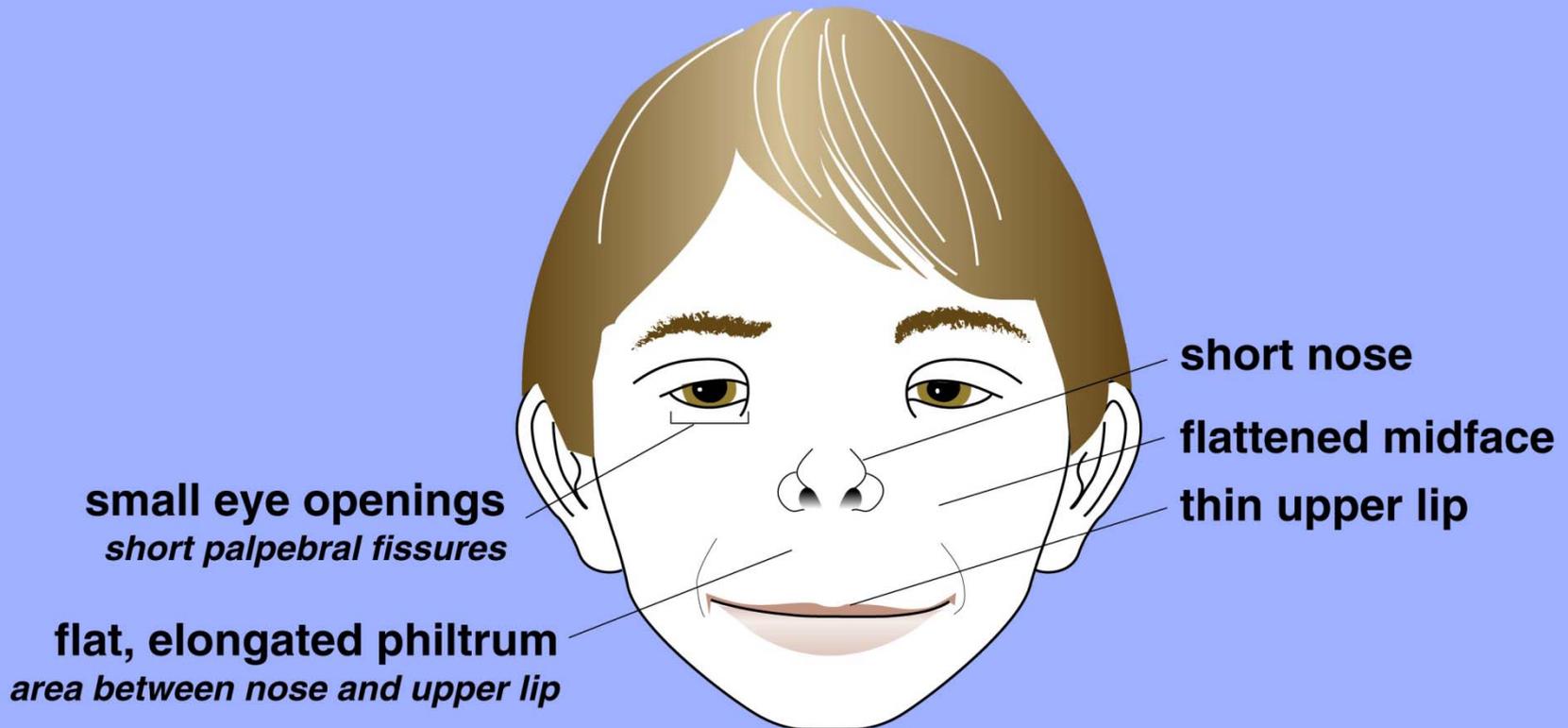
Photo: Fleming & Pfeifer, 2002



Facial features which are virtually constant in young children with FAS:

- Short palpebral fissures (horizontal measurement of the opening of the eyelids)
- Hypoplastic philtrum (smooth groove between the nose and mouth)
- Thin upper lip

Discriminating Features in FAS in Young Child



Short palpebral fissures, smooth philtrum, thin upper lip





FAS - changes in features in adolescence

- Facial appearance changes - nose and chin become more prominent but cardinal features persist (small palpebral fissures, smooth philtrum, thin upper lip)

(Clarren and Smith, NEJM, May, 1978)





FAS – Central Nervous System Effects

- ❑ Microcephaly (small head circumference)
- ❑ Structural brain malformations
- ❑ Intellectual disabilities
- ❑ Challenging behaviors



Diagnostic Assessment: Record Review/History

- Birth records (weight, length, head circ.)
- Growth records
- Medical records (birth defects?)
- IQ test results
- Neuropsychological testing or behavioral assessment



Asking about drinking behavior

- ❑ Birth mothers with active alcohol dependence
- ❑ Birth mothers in treatment
- ❑ Relatives of birth mother
- ❑ Foster parents
- ❑ Adoptive parents
- ❑ Other guardians



Asking about drinking behavior

- Birth mothers with active alcohol dependence
 - Denial
 - Frightened/defensive
 - Access (direct or indirect) to birth mother in a non-threatening environment
 - Empathy, nonjudgmental
 - Finding common ground
 - Work toward details
 - Healthy skepticism



Asking about drinking behavior

- Birth mothers in treatment
 - Empathy, nonjudgmental
 - Finding common ground
 - Work toward details
 - Usually most reliable information



Asking about drinking behavior

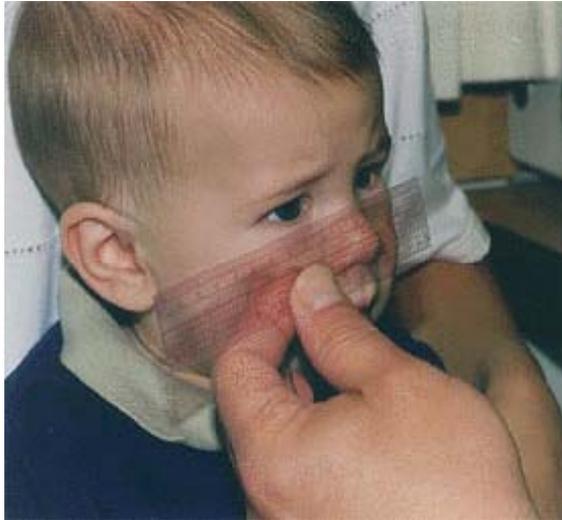
- Foster, adoptive families and other guardians
 - Clarify basis of information (direct observation vs. third hand report vs. supposition)
 - Finding common ground
 - Work toward details
 - Healthy skepticism (about accuracy, not intent)



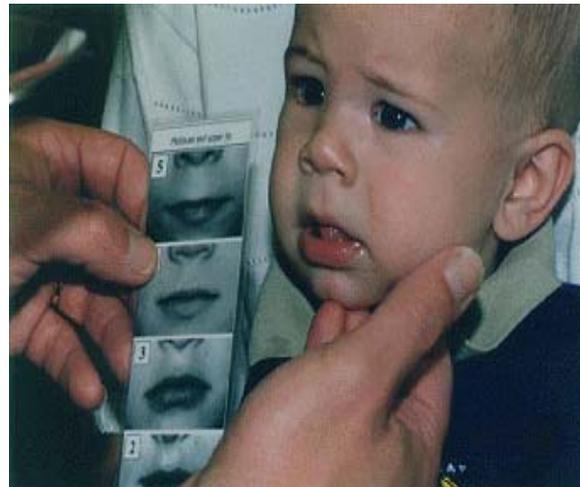
Physical assessment

- Growth measurements
- Palpebral fissure length
- Ears, midface, philtrum, lip (Washington card)
- Palate
- Heart
- Elbows, digits

Facial Assessment



Short Palbebral Fissures



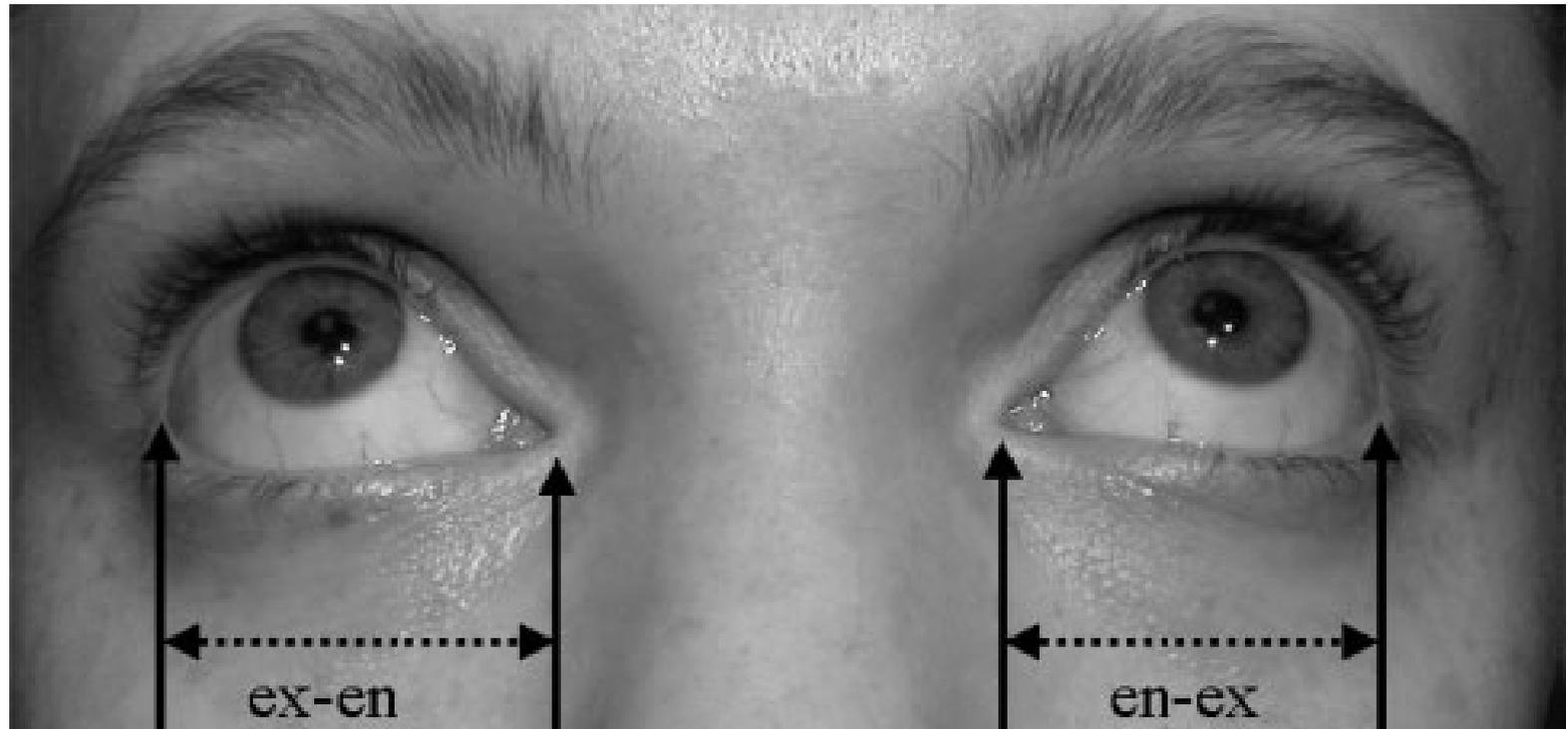
Thin Upper Vermillion



Indistinct Philtrum

Measuring palpebral fissures

(Chudley et. al., AJMG 145C August, 2007)



Differential diagnosis

- No single feature of FAS is unique to prenatal alcohol exposure.
- Other genetic conditions can cause growth and neurological impairments
- Other syndromes include a constellation of features *superficially* similar to FAS, including:
 - Williams Syndrome
 - Noonan Syndrome
 - Smith-Lemli-Opitz Syndrome
 - Fetal Hydantoin Syndrome

FASD Diagnostic Approaches

- Revised Institute of Medicine (“Hoyme”) criteria
 - Hoyme HE et.al., PEDIATRICS, 2005, v 115 #1, pp 39-47
- University of Washington diagnostic and Prevention Network 4-digit code
 - *Diagnostic Guide for FASDs*, Astley, 2004

IOM Diagnostic Categories

- ❑ **Fetal Alcohol Syndrome (FAS)**
- ❑ **Partial FAS:** some facial characteristics and at least *one* of the following—growth retardation, central nervous system impact, or behavior/cognitive impairments.
- ❑ **Alcohol Related Birth Defects (ARBD):** Congenital abnormalities known to be associated with prenatal alcohol exposure.
- ❑ **Alcohol Related Neurodevelopmental Disorder (ARND):** CNS deficits and/or behavioral/cognitive impairments known to be associated with prenatal alcohol exposure.

Institute of Medicine Diagnostic Categories and Criteria (Revised – “Hoyme” criteria)

- **FAS** with or without confirmed maternal alcohol exposure:
 - Characteristic minor facial anomalies
 - Pre- or postnatal growth impairment
 - Microcephaly or structural brain anomalies

Institute of Medicine Diagnostic Categories and Criteria (Revised – “Hoyme” criteria)

- **Partial FAS** with or without confirmed exposure
 - Characteristic facial features
 - Growth OR neurologic OR developmental impairments

Institute of Medicine Diagnostic Categories and Criteria

□ **Alcohol-related birth defects (ARBD)**

- Confirmed exposure
- Characteristic facial features
- Other major and/or minor anomalies

□ **Alcohol-related neurodevelopmental disorder (ARND)**

- Confirmed exposure
- Microcephaly OR structural CNS anomalies, OR
- Characteristic behavioral/cognitive abnormalities

University of Washington Diagnostic and Prevention Network 4-digit code

- *Diagnostic Guide for Fetal Alcohol Spectrum Disorders: The 4-Digit Code*
- 2004
- FAS Diagnostic and Prevention Network
- University of Washington



4-Digit Code

- Digits reflect magnitude of expression
- 256 Possible 4-Digit Codes
- 22 Unique Clinical Diagnostic Categories
- System does not use ARND
- Head Circumference at/below 3rd percentile (vs. 10th)
- Palpebral Fissure Length at/below 3% (vs. 10th)

4-Digit Code: Example 3444: “FAS-Alc Exposed”

Significant	Severe	Definite	4		X	X		X	4	High Risk
Moderate	Moderate	Probable	3	X					3	Some Risk
Mild	Mild	Possible	2						2	Unknown
None	None	Unlikely	1						1	No Risk
Growth Deficiency	FAS Facial Features	CNS Damage		Growth	Face	CNS		Alcohol		Prenatal Alcohol



Diagnostic Categories

- Sentinel Physical Findings (growth and/or facial features)
- Static Encephalopathy (brain structure abnormalities)
- Neurobehavioral Disorder (cognitive/behavioral w/o structural abnormalities)
- Alcohol Exposure
- FAS (with or w/o known exposure)
- Partial FAS
- FAS Phenotype (no exposure)

General challenges to diagnosis of FASD's

- Awareness among physicians and service providers
 - Of disorder and *spectrum* of effects
 - Of exposure history
 - Of availability of diagnostic services
 - Of availability of management services
- Willingness to refer for assessment

General challenges to diagnosis of FASD's

- Lack of reliable information regarding exposure
- Biases (either direction) in exposure information
- Variables that affect phenotype:
 - Timing of exposure and dosage
 - Maternal “susceptibility” (Genetic? Metabolic?)
 - Embryonic/fetal “susceptibility” (Genetic? Metabolic?)
 - Others unknown...

General challenges to diagnosis of FASD's

- Availability of diagnostic services, especially optimal multidisciplinary setting
- Difficulty obtaining health and growth records
- Standardization of the examination
 - Measurement techniques
 - Standards used
 - Cutoff definitions of normal and abnormal
 - Adjustments for race??



Challenges in adolescents and adults

- ❑ Greater difficulty getting accurate exposure information, birth records, growth records, medical records
- ❑ Morphologic changes (next)
- ❑ Inadequate mechanisms for long term follow up and management
- ❑ Adults who were assessed as children may not be aware of that, or of how/where diagnosis was made



Framework for FAS Diagnosis and Services – essential elements

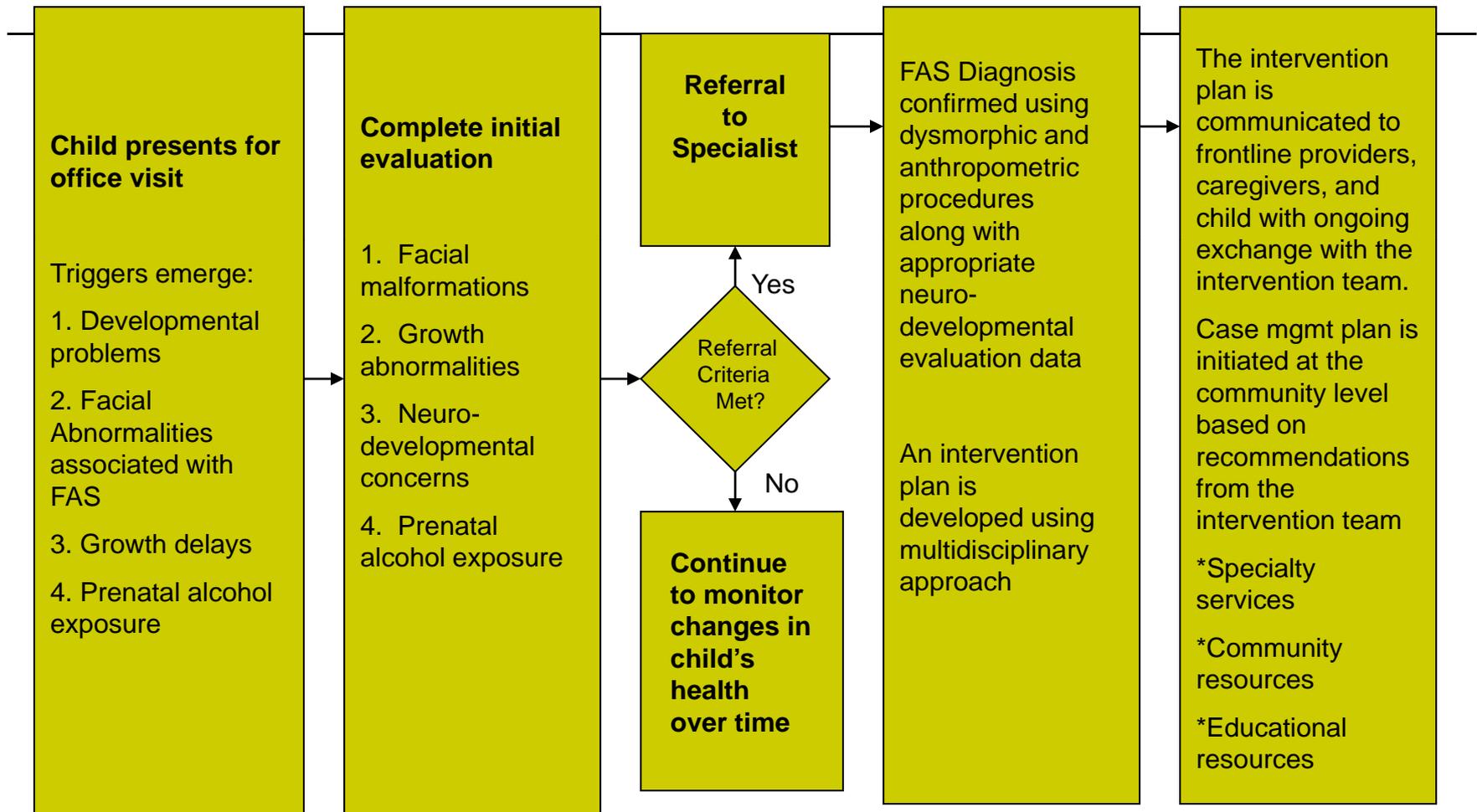
- Initial suspicion/identification
- Referral
- Diagnosis
- Services

FAS Diagnostic Framework

Provider Contact

Diagnosis

Services





American Academy of Pediatrics

- Expert panel working with CDC and RTC's to provide pediatricians with practical information and guidelines for each step

Initial Identification:

What triggers a referral for assessment?

- Developmental or behavioral concerns
- Facial characteristics associated with FAS
- Growth deficits
- Maternal alcohol use
 - Other exposures associated with alcohol use (e.g. cocaine)
 - Adoption from endemic region
- Population screening?
- Do consistent standards or protocols exist? (hint: not really, not yet)



Initial identification – variables & barriers

- Awareness of FASDs
- Acceptance of the existence of FASDs
- Asking, why? (parents, providers)
- Having other explanations (birth injury, cerebral palsy, ADHD, family background, etc.)



Referral: *What drives the referral?*

- Need for answers (parents, physician, patient)
- Need for services
 - Financial (Social security, subsidized adoption)
 - Educational (IEP, speech/language, behavior)
 - Physical (OT/PT, therapeutic programs)
 - Vocational (Division of Vocational Rehabilitation)
 - Legal
 - Medical (heart defects, cleft palate, seizures, behavior)



Referral – variables & barriers

- ❑ Awareness of FASDs
- ❑ Acceptance of the existence of FASDs
- ❑ Concerns about stigmatization of parent, child
- ❑ Guilt/blame
- ❑ Acknowledgement that there is benefit to having a diagnosis
- ❑ Provider familiarity/confidence with diagnostic skills, availability of resources

Diagnosis

- Where do we refer for diagnostic assessment?
 - Multidisciplinary FASD clinic
 - Genetics clinic – “dysmorphology”
 - Pediatrics clinic
 - Other – child development, neurology, psychiatry
- What options for diagnostic evaluation currently exist in your community? Within a reasonable referral distance?

Diagnosis

- What diagnostic criteria should be used?
 - Facial features
 - Smooth philtrum
 - Thin vermilion (pink portion) of upper lip
 - Small palpebral fissures
 - Growth deficits
 - Central nervous system abnormality
 - Confirmation of exposure?

Diagnosis

- How do we measure these criteria?
 - Techniques
 - Normal standards
 - Use consistent percentiles to define normal ranges (<10th, 3rd? 5th?)
 - (more in breakout session)

Diagnosis

- What diagnostic schema should be used?
 - CDC Guidelines (for FAS)
 - “Hoyme” criteria
 - American Academy of Pediatrics
 - University of Washington 4-Digit Code
 - National Institutes of Health
 - Other?



Goals of the Assessment

- What does the family need to learn?
- Assess strengths and deficits
- Assess interactions and relationships
 - How are sociocultural factors influencing the patient, family
- Specific, concrete, viable recommendations
 - At diagnosis, or subsequent referral(s)?

Services

- Where do we refer?
 - What services have already been accessed?
 - Are “FASD-specific” services required?
 - Should clinicians have specific training?
 - Will services for individuals with developmental disabilities (i.e. Down syndrome, autism, ADHD) serve the needs of children with FASDs?



Who should be on the 'team'

- Ideal team members?
 - Physician
 - Occupational Therapist
 - Physical Therapist
 - Nutritionist
 - Psychologist
 - Audiologist
 - Speech and Language Pathologist
 - Social Worker

- What is realistic for your community?
 - Most communities cannot support a multidisciplinary team

All Communities

- All communities should develop a ‘framework’
- No two will be identical
- Complete lack of services is *exceedingly rare*
 - Identify what is available
 - Provide training to clinicians
 - Modify to FAS as appropriate