Fetal Alcohol Spectrum Disorders

Cynthia C. Mondesir, MD

Fetal Alcohol Syndrome Disorders

- **Fetal Alcohol Syndrome –** Fetal Alcohol Syndrome is a clinical diagnosis for those exposed to alcohol prenatally showing growth retardation, facial features that are characteristic of FASD, and central nervous system problems.
- **Alcohol Related Neurodevelopmental Disorder –** individuals with a confirmed maternal alcohol use, neurodevelopmental abnormalities, and unexpected slow development.
- **Alcohol Related Birth Defects –** individuals with confirmed maternal alcohol use and one or more congenital defects which includes the heart, bone, kidney, vision or hearing abnormalities.

Fetal Alcohol Spectrum Disorders

- CDC studies have identified 0.2 to 1.5 infants with FAS for every 1,000 live births in certain areas of the United States.[1]
- The most recent CDC study analyzed medical and other records and found FAS in 0.3 out of 1,000 children from 7 to 9 years of age.[2]

1 CDC. Fetal alcohol syndrome-Alaska, Arizona, Colorado, and New York, 1995-1997. MMWR Morb Mortal Wkly Rep. 2002;51(20):433-5. [Read article]

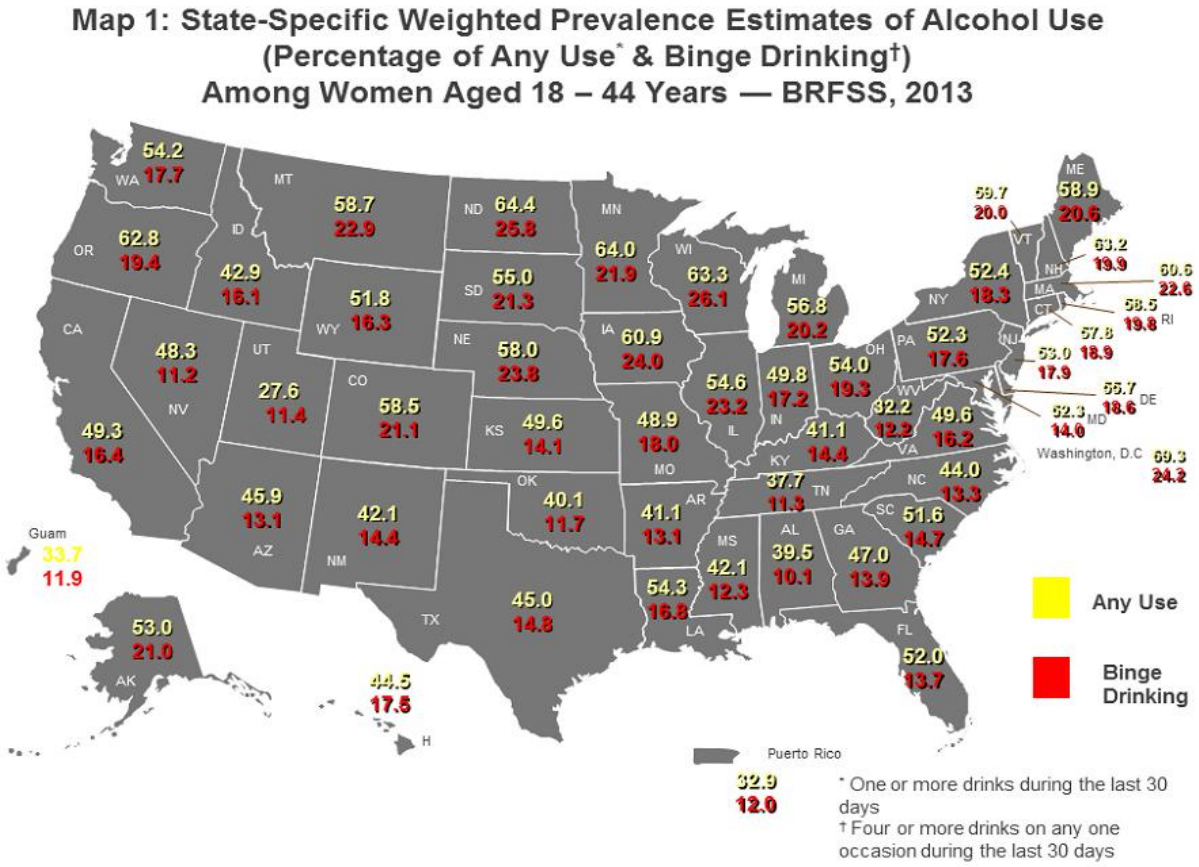
2 CDC. Fetal Alcohol Syndrome Among Children Aged 7-9 Years – Arizona, Colorado, and New York, 2010. MMWR Morb Mortal Wkly Rep. 2015;64(3):54-57. [Read article]

Fetal Alcohol Spectrum Disorders

- Community studies using physical examinations, experts estimate that the full range of FASDs in the United States and some Western European countries might number as high as 2 to 5 per 100 school children (or 2% to 5% of the population)[3]

3.May PA, Baete A, Russo J, Elliott AJ, Blankenship J, Kalberg WO, Buckley D, Brooks M, Hasken J, Abdul-Rahman O, Adam MP, Robinson LK, Manning M, Hoyme HE. Prevalence and characteristics of fetal alcohol spectrum disorders. Pediatrics. 2014;134:855-66.

Alcohol Use and Binge Drinking



Alcohol use and binge drinking among women of childbearing age – United States, 2011–2013Morbidity and Mortality Weekly Report; September 25, 2015; 64(37); 1042-1046 CDC

Data are from the Behavioral Risk Factor Surveillance System (BRFSS): This telephone survey tracks national and state-specific health risk behaviors of adults, aged 18 years and older, in the United States.

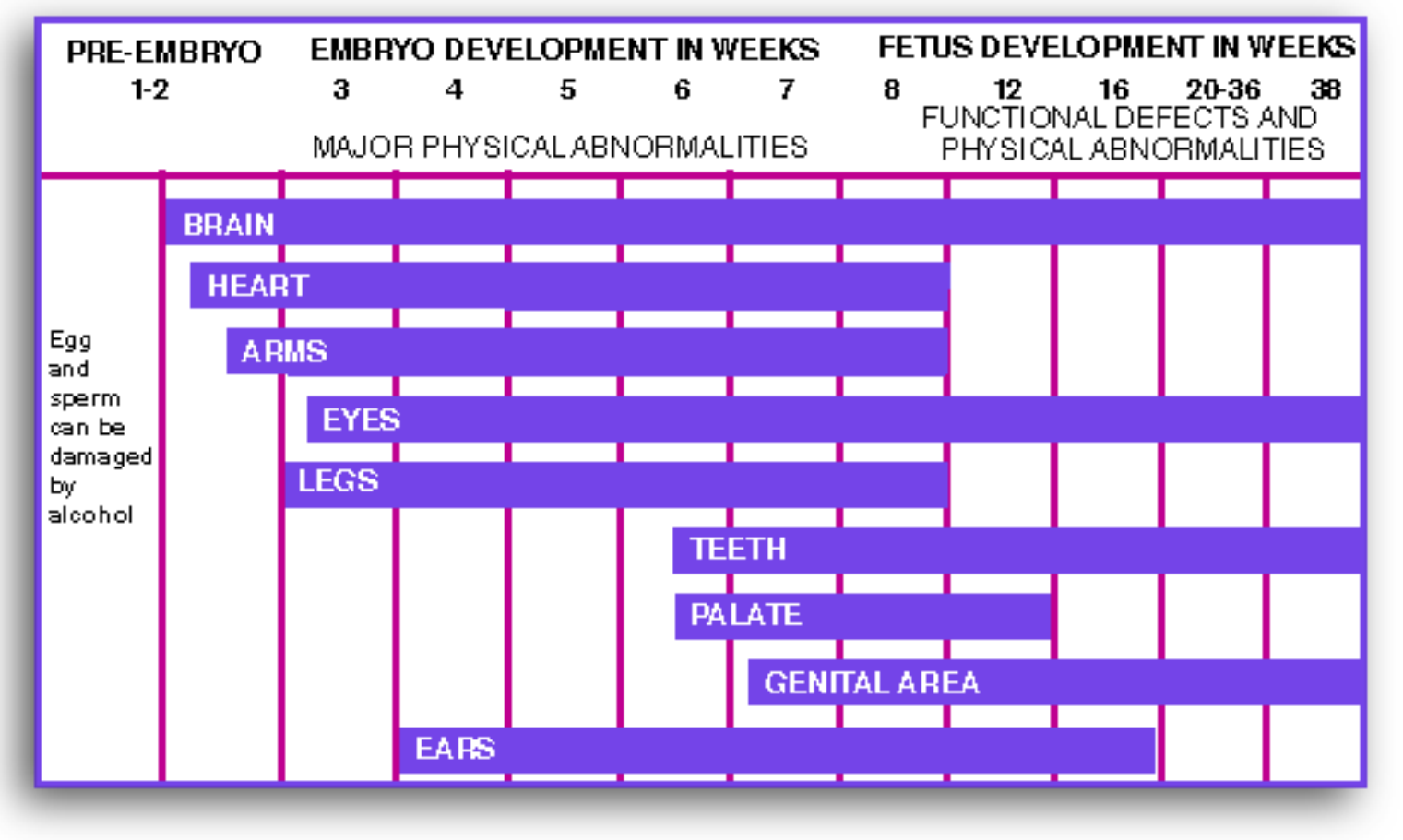
Fetal Alcohol Spectrum Disorders

- According to recent State of Alaska DHSS surveillance data, more than **126** children are born at risk for FASD each year in Alaska.
- Alaska reports the highest documented prevalence of FASD in the United States
- FASD is preventable.

Costs

- The Alaska Department of Health and Social Services
  - each child born with FASD in Alaska will cost the State of Alaska between $860,000 and $4.2 million dollars from birth to age 18.
  - The effects of FASD can be seen throughout the education, health, social service, labor, and corrections systems of the Alaska State Government.

Alcohol is a powerful Teratogen



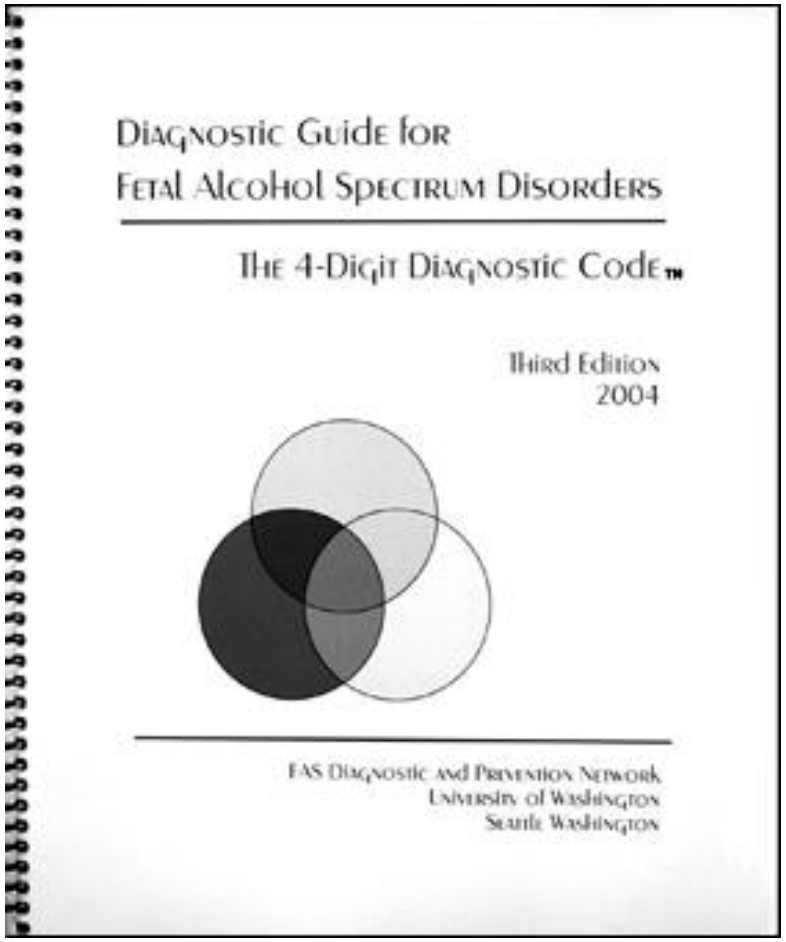
Source: http://www.thinksciencemaurer.com/wp-content/uploads/2015/05/FASbaby1.gif

Primary Diagnostic Criteria

- Growth deficiencies -stunted prenatal and/or postnatal growth.
- Permanent brain damage resulting in neurological abnormalities, delay in development, intellectual impairment and learning/behavior disabilities.
- Abnormal facial features including short eye opening, short nose, flat midface, thin upper lip and small chin.
- Maternal alcohol use during pregnancy. The window of exposure for alcohol is the entire 9 months of pregnancy. Whatever is developing, at the time of exposure to the fetus, can be affected.

Sterling K. Clarren, M.D. and Susan J. Astley, Ph.D., Diagnostic Guide for Fetal Alcohol Spectrum Disorders and Related Conditions, 1997

Diagnostic Guide

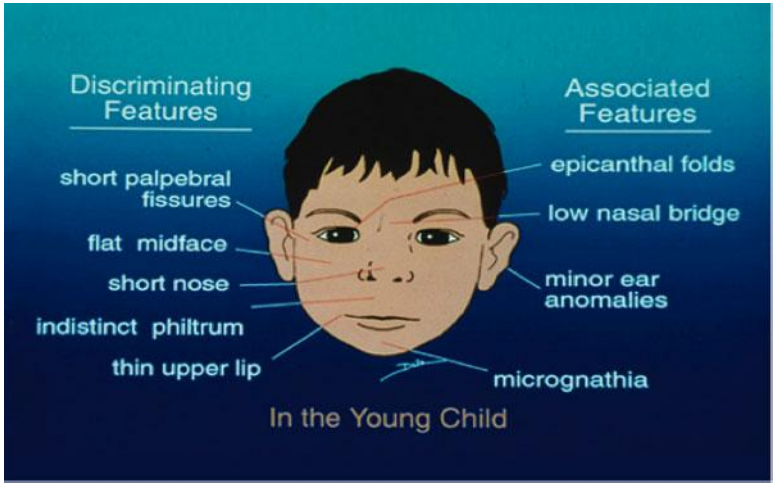


Growth

- Children with FAS have height, weight, or both that are lower than normal (at or below the 10th percentile).
- These growth issues might occur even before birth. For some children with FAS, growth problems resolve themselves early in life.

Facial Features

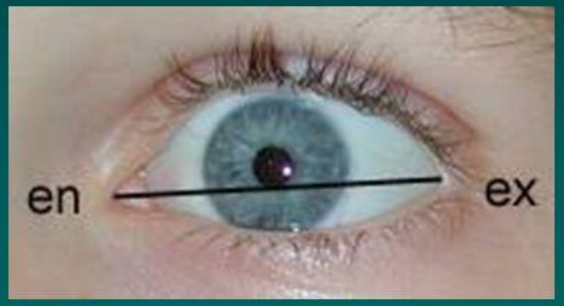
- Smooth ridge between the nose and upper lip (smooth philtrum)
- Thin upper lip
- Short distance between the inner and outer corners of the eyes, giving the eyes a wide-spaced appearance.



Face

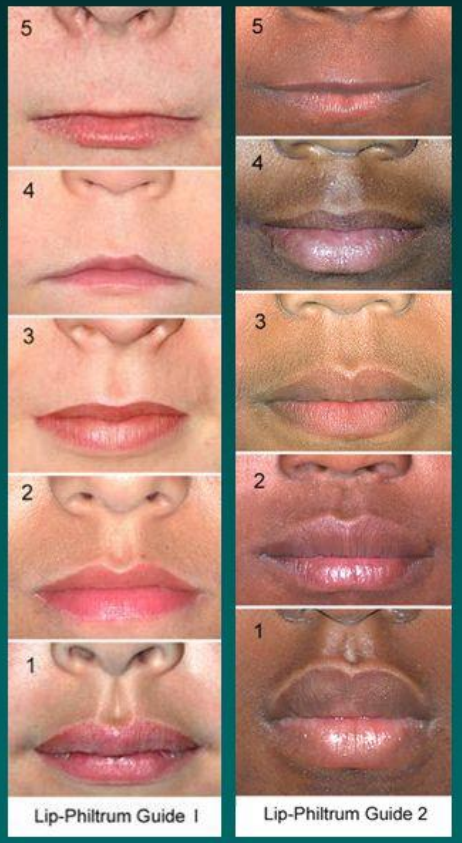
The Three Diagnostic Facial Features of FAS

1)	Short PFL	<= -2 SD
2)	Smooth Philtrum	Rank 4 or 5
3)	Thin Upper Lip	Rank 4 or 5



Palpebral fissure length (PFL) = endoncanthion to exocanthion

FAS



CNS Structural

- Smaller-than-normal head size for the person's overall height and weight (at or below the 10th percentile).

- Significant changes in the structure of the brain as seen on brain scans such as MRIs or CT scans.

CNS Neurologic

- There are problems with the nervous system that cannot be linked to another cause.
- Examples include poor coordination, poor muscle control, and problems with sucking as a baby.

CNS Function

- The person's ability to function is well below what's expected for his or her age, schooling, or circumstances.

  - ✓ **Cognitive deficits** (e.g., low IQ), or significant developmental delay in children who are too young for an IQ assessment.

Or

Problems in at least three of the following areas:

- ✓ **Cognitive deficits** (e.g., low IQ) or developmental delays.

  Examples include specific learning disabilities (especially math), poor grades in school, performance differences between verbal and nonverbal skills, and slowed movements or reactions.

CNS Function

- **Executive functioning deficits.**

  These deficits involve the thinking processes that help a person manage life tasks. Such deficits include poor organization and planning, lack of inhibition, difficulty grasping cause and effect, difficulty following multistep directions, difficulty doing things in a new way or thinking of things in a new way, poor judgment, and inability to apply knowledge to new situations.

- **Attention problems or hyperactivity**

  A child with these problems might be described as "busy," overly active, inattentive, easily distracted, or having difficulty calming down, completing tasks, or moving from one activity to the next. Parents might report that their child's attention changes from day to day (e.g., "on" and "off" days).

- **Motor functioning delays**

  These delays affect how a person controls his or her muscles.

  Examples include delay in walking (gross motor skills), difficulty writing or drawing (fine motor skills), clumsiness, balance problems, tremors, difficulty coordinating hands and fingers (dexterity), and poor sucking in babies

CNS Function

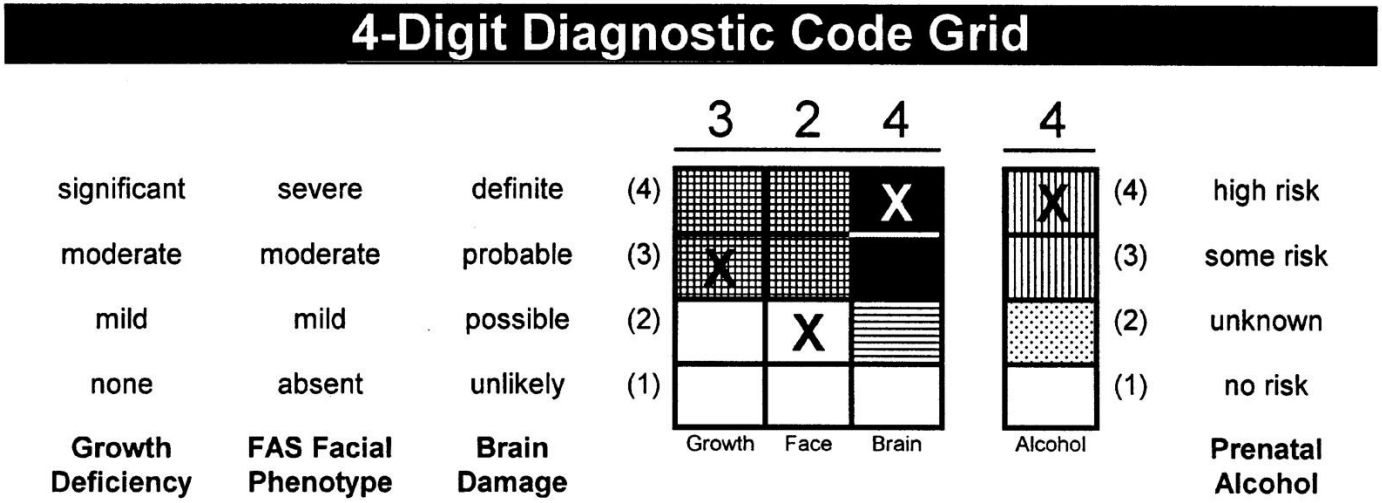
- **Problems with social skills**
  - A child with social skills problems might lack a fear of strangers, be easily taken advantage of, prefer younger friends, be immature, show inappropriate sexual behaviors, and have trouble understanding how others feel.

- **Other problems**
  - can include sensitivity to taste or touch, difficulty reading facial expression, and difficulty responding appropriately to common parenting practices (e.g., not understanding cause-and-effect discipline)

Maternal Alcohol Use

- Confirmed alcohol use during pregnancy can strengthen the case for FAS diagnosis. Confirmed absence of alcohol exposure would rule out the FAS diagnosis. It's helpful to know whether or not the person's mother drank alcohol during pregnancy.
- But confirmed alcohol use during pregnancy is not needed if the child meets the other criteria.

Diagnostic Code



Bethel FASD Diagnostic Team

- Pauline Thomas, Case Manager/FASD Coordinator
- Dr. Cynthia Mondesir, Pediatrician
- Dr. Mien Chyi, Pediatrician
- Dr. Sarah Angstman- Pediatric Psychologist
- Kiera Gefroh, Physical Therapist
- Monica Leinberger, Parent Navigator, Guardian ad litem AVCP

Case Presentation

- 9 y/o male with h/o developmental delay, seizure disorder and VSD
- Maternal aunt describes him as having poor impulse control, gets in trouble a lot at school, lashes out at other children when he is being teased. They wish to have him evaluated obtain services.
- Adopted by maternal grandfather
- Mother drank ETOH until the 5th month of pregnancy

Case Presentation

- Child is at 90-95% for height and weight
- Palpebral Fissure < -2SD,
- Philthrum 3
- Upper Lip Thinness 1 or 2

Facial Rank

A

5-point Likert Rank for philtrum and lip	*z* score[a] for palpebral fissure length	Circle the ABC scores for: Palpebral fissure length	Philtrum smoothness	Upper-lip thinness
4 or 5	≤ −2 SD	**C**	**C**	**C**
3	> −2 SD and ≤ −1 SD	**B**	**B**	**B**
1 or 2	> −1 SD	**A**	**A**	**A**

B

4-digit facial rank	Level of expression of FAS facial features	Palpebral fissure: philtrum-lip ABC-score combinations
4	Severe	CCC
3	Moderate	CCB, CBC, BCC
2	Mild	CCA, CAC, CBB, CBA, CAB, CAA BCB, BCA, BBC, BAC ACC, ACB, ACA, ABC, AAC
1	None	BBB, BBA, BAB, BAA ABB, ABA, AAB, AAA

Neuropsychiatric Evaluation

ASSESSMENT PROCEDURES

Clinical Interview and Mental Status Exam (8/17/16)
Wechsler Intelligence Scale for Children-V (WISC-V; 8/17/16)
Wide Range Achievement Test-4 (WRAT-4; 8/17/16)
California Verbal Learning Test- Children's Version (CVLT-C; 8/17/16)
Conner's 3 Parent Form (Josephine Hoelscher; 8/17/16)
Adaptive Behavior Assessment System-3 (Josephine Hoelscher; 8/17/16)
Review of YKHC Record (8/24/16)
Review of LYSD Special Education records (Received 8/22/16)
Review of ANMC records (Received 8/17/16)
Interview with Edgar Hoelscher (8/26/16)

Neuropsychiatric Evaluation

Patient is experiencing problems in his verbal comprehension and language based academic achievement

Scores fall below the expected level in working memory, processing speed, auditory attention and non verbal reasoning.

4 Digit Score

- Growth 1
- Face 2
- CNS 4
- Alcohol 4
- DX: Static Encephalopathy Alcohol Exposed

What Next?

- PREVENTION
- Identify infants and children who may have been exposed to alcohol in utero
- Provide early services and protective environments to prevent the long term disabilities of FASD

References

- University of Washington FASD Diagnostic and Prevention Network

  http://depts.washington.edu/fasdpn/
- www.cdc.gov
- Alaska Department of Health and Social Services

  http://dhss.alaska.gov/dbh/fas/Pages/default.aspx