Fetal Alcohol Spectrum Disorders: An Overview

Midwest Regional FAS Training Center
MRFASTC
History of FASD

- “Behold, thou shalt conceive, and bear a son: and now drink no wine or strong drink.” - Judges 13:7

- “Foolish, drunken and harebrained women, most often bring forth like unto themselves, morose and languid.” – Aristotle

- “The fatal effects of the frequent use of distilled spirituous liquors…and too often the cause of weak, feeble and distempered children.” – The British College of Physicians (1725)
History, continued

- Dr. William Sullivan, in 1899, compared cohorts of female alcoholic prisoners to blood relatives, married to sober husbands.
  - Infant mortality among alcoholic prisoners was 44%, but only 24% in their blood relatives.
  - Women, who entered prison early in their pregnancies delivered healthier infants, than women who entered prison later in their pregnancies
- Dr. David Smith and Dr. Ken Jones, in 1973, created the acronym, Fetal Alcohol Syndrome
  - Then, Fetal Alcohol Effect, Partial FAS, Alcohol Related Birth Defects, Alcohol Related Neurodevelopmental Disorder.
What is Fetal Alcohol Spectrum Disorder (FASD)?

- Umbrella term describing the range of effects that can occur in an individual whose mother drank alcohol during pregnancy.
- Includes physical, mental, behavioral, and/or learning disabilities, with possible lifelong implications.
- Not a clinical diagnosis.
- Includes fetal alcohol syndrome (FAS), as well as less global effects of alcohol upon a fetus [alcohol-related neurodevelopmental disorder (ARND) and alcohol-related birth defects (ARBD)].

Fetal Alcohol Syndrome Recognition

“Fetal alcohol syndrome (FAS) now is recognized as the leading known cause of mental retardation in the United States, surpassing spina bifida and Down’s syndrome.” - JAMA, 1991
**Incidence of Birth Defects**

- Down syndrome ............... 1/800 births
- Cleft lip +/- palate ............ 1/800 births
- Spina bifida .................... 1/1000 births
- Fetal alcohol syndrome ..... 1-2/1000 births
  - ~61600 U.S. babies with FAS per year
  - Many more with alcohol induced problems
  - Higher rates in some populations
  - CDC data, literature review
- **New research indicates rates of 2-7/1000 births**
  - 2-5% of younger school age children

FASD and AZ  (2008 statistics according to www.fasarizona.com)

- Of 103,803 babies born in 2007, estimated 12.4% (more than 12,000 babies) were born to mothers who engaged in binge drinking during pregnancy.
- Approx 60,000 individuals are living with a significant degree of FASD, and at least 6,000 may have full FAS.
- Cost to AZ taxpayers estimated to be $98 million/year; Annual cost just for Special education and Juvenile Justice in AZ due to FASD each year is approx $27 million.
Cost to Society

• Health care costs for persons with FASD in 2003: $5.4 billion
• Lifetime health care costs per person with FASD: $860,000 to $4.2 million

• Very conservative estimate of costs.
• Includes medical care and lost productivity.
• Does not include mental health care, special education, foster care, supported living or employment, costs associated with criminal justice.
### Categories of Prenatal Alcohol Damage
(adapted from Stratton, Howe, & Battaglia (1996), Institute of Medicine)

<table>
<thead>
<tr>
<th>Category</th>
<th>Description</th>
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<tbody>
<tr>
<td><strong>Category 1 (FAS with confirmed maternal alcohol exposure)</strong></td>
<td>Utilizes full criteria for FAS</td>
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<tr>
<td><strong>Category 2 (FAS without confirmed maternal alcohol exposure)</strong></td>
<td>Utilizes full criteria for FAS but alcohol exposure cannot be confirmed</td>
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<tr>
<td><strong>Category 3 (Partial FAS with confirmed maternal alcohol exposure)</strong></td>
<td>Some facial features of FAS; Evidence of either growth deficiency, CNS neurodevelopmental abnormalities, or a complex pattern of cognitive and behavioral abnormalities; confirmed alcohol exposure in utero</td>
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<tr>
<td><strong>Category 4 (Alcohol related birth defects [ARBD])</strong></td>
<td>Confirmed alcohol exposure in utero; Evidence of physical anomalies that have been linked to maternal ingestion of alcohol</td>
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<tr>
<td><strong>Category 5 (Alcohol related neurodevelopmental disorder [ARND])</strong></td>
<td>Confirmed alcohol exposure in utero; No physical manifestations of FAS but do have CNS neurodevelopmental abnormalities related to prenatal alcohol exposure</td>
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Diagnosis of FAS: CDC Criteria

FAS diagnosis requires **all three** of the elements below.

Documentation of:

1. **All three** facial abnormalities – smooth philtrum, thin upper lip, small palpebral fissures
2. Growth deficits
3. Central nervous system/neurobehavioral deficits (including structural, neurological, and/or functional)
   1. r/o other possible diagnoses

**Confirmed prenatal alcohol use can strengthen the evidence for diagnosis, but is not necessary in the presence of all findings listed above.**

(CDC Health Provider’s Toolkit, 2005)
Facial Features of FAS

Discriminating Features
- short palpebral fissures
- flat midface
- short nose
- indistinct philtrum
- thin upper lip

Associated Features
- epicanthal folds
- low nasal bridge
- minor ear anomalies

In the Young Child
CDC Criteria (2004)

- Facial Abnormalities
  - **Smooth philtrum**
    - Lip philtrum guide 4 or 5
  - **Thin vermillion**
    - Lip philtrum guide 4 or 5
  - **Palpebral fissures**
    - 10th percentile or lower
Source: Wattendorf, DJ, & Muenke, MD (2005), American Family Physician, 72(2) 279-285.
Faces of FAS*
Growth in FAS

- Weight
- Length

↑ Birth  [Normal]  [FAS]
CDC Criteria (2004)

- Central Nervous System or Neurobehavioral Disorders
  - Structural
    - Cell death and incorrect migration
    - Nerve formation
    - Regional brain development failures
    - Decrease in CNS regeneration
  - Neurological effects
  - Functional effects
Diagnostic Pearls

- Best time: 8 months to 8 years
- Best person: Pediatric Dysmorphologist or Geneticist
- Differential diagnosis
  - Williams syndrome (ELN deletion)
  - Velocardiofacial syndrome (del 22q11)
  - Noonan syndrome (PTPN deletion)
  - DeLange syndrome
  - Dubowitz syndrome
  - Maternal PKU embryopathy
  - Maternal Toluene embryopathy
How Common is Alcohol Use Among Women?
Alcohol Consumption Among Women

Categories of Alcohol Consumption

- Any
- Frequent
- Binge

Percent Women

- Might become pregnant
- Pregnant

Any: 54.9%
Frequent: 10.1%, 1.9%
Binge: 12.4%, 1.9%
Alcohol Consumption is Higher in Early Pregnancy

- 45% of women reported consuming alcohol in the three months before finding out they were pregnant.
- 2/3 of women did not learn they were pregnant until the 4th – 6th week of gestation.
- CDC encourages a pre-conceptual approach to alcohol screening.
Who is at highest risk for having a child with FASD?

- Having had a child with FAS
- History of binge drinking
- Long history of drinking
- **Any Woman Who Drinks During Pregnancy**
Risks of FASD Among Women Who Drink During Pregnancy

- **Heavy/Binge** (>3 per occasion)
- **Daily/Almost Daily** (<3 per occasion, >24 occasions/month)

Risk in those not in the 2 categories on the graph = 0%
Alcohol Metabolism and Pharmacology

- Absorption
- Distribution
- Metabolism and elimination
Absorption

- Woman takes a drink → introduced into the stomach → absorption/metabolism of the molecule (C2H5OH) occurs rapidly
- Peak blood ethanol concentrations attained approx 1 hour after consumption
- Women attain consistently higher blood ethanol concentrations than men following equivalent amounts of ethanol consumption
Distribution

- Compartmentalization
  - Because of alcohol’s rapid solubility in water, it can easily cross cell membranes into the cell (which is 98% water)
  - Alcohol is less soluble into lipids and compartments with substantial lipids
- Placental effects and fetal distribution
  - The placenta acts as a selective barrier
  - Alcohol is easily passed by diffusion from the maternal blood into the fetal blood
Genetic or polymorphic variants of the ALDH gene might influence ethanol metabolism in some populations.

The placenta does not metabolize ethanol well; the capacity for ethanol metabolism by the embryo/fetus increases with gestational age.

An embryo or early fetus lacks the enzymes for metabolism, thus the mother must metabolize most of the alcohol. Removal of the alcohol in the embryo/fetus occurs by simple diffusion back to the mother.

Embryonic alcohol levels might be higher in the embryo than in the mother and be present for a more prolonged, variable time due to limited embryonic metabolism.
Effects of Alcohol on the Fetus

- Miscarriage
- Premature Birth
- Low Birth Weight
- Development Complications
- FASD
Effects of Alcohol on the Fetus

- Central Nervous System
- Craniofacial defects
- Growth retardation
- Cardiovascular defects
- Renal defects
- Skeletal defects
Fetal Exposure

- Decreased blood-brain barrier
- Dermal Exposure
- Inhaled Exposure
- Amniotic ETOH Levels
- Maternal BAL
- Placental BAL
Mean ± S.D. ethanol concentration – time curves in maternal blood and amniotic fluid of six healthy pregnant woman at 16-18 weeks of gestation. Data were obtained from Brien et al. [5].
When is the embryo or fetus most susceptible to alcohol exposure?
Development of the Embryo

- **Central Nervous System (CNS)**
  - Heart
  - Arms
  - Eyes
  - Legs
  - Teeth
  - Palate
  - External Genitalia
  - Ears

- **Period of the Ovum**
  - 1-2

- **Period of the Embryo (in weeks)**
  - 3
  - 4
  - 5
  - 6
  - 7
  - 8

- **Period of the Fetus (in weeks)**
  - 12
  - 15
  - 20-36
  - 38

- **Most Common Site of Birth Defect**
Alcohol Effects on Neurons

What are primary mechanisms by which alcohol damages the brain cells during development?
Neuronal Responses to Alcohol Exposure

- Neurogenesis – neuron ‘birth’
- Migration
- Growth & Differentiation
- Synaptogenesis
- Apoptosis
- Plasticity
Brain Damage From *In Utero* Alcohol Exposure
FAS and the brain

Child’s Brain Unexposed to Alcohol

- Fewer folds in brain
- Smaller head size
- Flattened face

Child’s Brain after Prenatal Exposure to Alcohol

- Smoother surface of brain
- Underdeveloped inner structure of brain

Source: Children’s Research Triangle, Chicago, IL
### Brain Structures Most Sensitive to Prenatal Alcohol Exposure

**Unaffected Brain**
- **Corpus Callosum**
- **Cerebellum**

**Affected Brain**
- Overall smaller and less developed brain

<table>
<thead>
<tr>
<th>Brain Structure</th>
<th>Function</th>
<th>Prenatal alcohol exposure may result in problems with:</th>
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<tbody>
<tr>
<td>Corpus Callosum</td>
<td>Communicates motor, sensory and cognitive information between the two hemispheres of the brain</td>
<td>Storing and retrieving information, problem solving, attention and verbal memory</td>
</tr>
<tr>
<td>Cerebellum</td>
<td>Processes input from other areas of the brain to coordinate motor and cognitive skills</td>
<td>Controlling movements, maintaining balance and fine motor skills</td>
</tr>
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National Organization on Fetal Alcohol Syndrome (NOFAS)
1.800.66NOFAS or visit www.nofas.org
Functional MRI and FASD*

Children and adults with FAS had decreased activity in areas associated with executive function, working memory, and attention.

*K. Maliza, et. al., Functional MRI of Fetal Alcohol Syndrome
MRI Findings in FASD*

- Decrease in white matter in temporal and parietal lobes.
  - Associational, learning.
- Decreased size of frontal lobes
  - Problems with response inhibition, behavioral control, executive function.
- Cerebellum size decreased up to 15%
  - Muscular coordination
- Corpus Callosum smaller or absent
  - Complex thinking
- Basal Ganglia
  - Posture and movement. Dopamine.

*Edward Riley and Christie McGee, San Diego State University
Low- Level Drinking

- Low levels of alcohol exposure have been linked to fetal alcohol effects
  - Neuro-behavioral and cognitive effects can be seen without associated facial features
  - Alterations in dopamine system in the brain: both development and life-long function proven in monkey studies
Prenatal Alcohol Exposure

What happens to the brain…

- Graphomotor skills
- Attachment
- Self control and regulation
- Memory, recall and learning
- Abstraction
- Mood regulation

- General IQ
- Attention
- Social communication
- Social interaction skills
- Executive functioning
- Math
FAS and the Brain—IQ at 8 years

PEA = prenatal exposure to alcohol; NC = normal control; FSIQ = full scale IQ; VIQ = verbal IQ; PIQ = performance IQ

Mattson: J Pediatrics, Volume 131(5), November 1997; 718-721
Cognitive and Behavioral Impairments

- Memory
- Attention
- Learning
  - Verbal
  - Visual-Spatial
- Reaction Time
- Executive Functions
Attention

- Hallmark of PAE
- Research has consistently reported attentional deficits on tasks of vigilance and reaction time and parent/teacher measures
- PAE linked to slower overall processing speed, particularly on more effortful tasks
EF

- Overall, both cognition based and emotion related aspects of EF appear to be impaired in persons with FASD
- BRIEF – deficits on BRI and MI
- Tower tasks – deficits in planning and problem solving
- Verbal fluency – more pronounced impairments on letter fluency tasks
Inconsistent findings, but generally have been found to score lower on measures of language functioning.

FASD and NEPSY-II – children and adolescents with FASD demonstrated significantly lower performance than controls on measure of oral language comprehension on NEPSY-II.
Visual Spatial

- Often perform poorly on tasks that involve learning spatial relationships among objects
Quantitative Reasoning

- Consistently documented as deficit in PAE
- Greater deficits on measures of mathematical and arithmetic ability compared to controls
- Degree of impairment is strongly correlated with degree of alcohol exposure in utero
- Calculation and estimation more impaired
Learning and Memory

- Poorer on verbal memory, particularly during early stages of memory formation (i.e. encoding)
- Impaired visual memory abilities, particularly for faces
Social Cognition

- Impairments in social functioning
## Prenatal Alcohol Exposure

<table>
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<tr>
<th>Cognitive Deficit</th>
<th>Behavioral Manifestation</th>
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<tbody>
<tr>
<td>Poor judgment</td>
<td>Easily victimized</td>
</tr>
<tr>
<td>Attention deficits</td>
<td>Distractible, impulsive</td>
</tr>
<tr>
<td>Arithmetic disability</td>
<td>Poor finances</td>
</tr>
<tr>
<td>Memory impairment</td>
<td>Poor learning</td>
</tr>
<tr>
<td>Difficulty abstracting</td>
<td>Poor understanding of consequences</td>
</tr>
<tr>
<td>Disorientation in space/time</td>
<td>Poor social cue recognition</td>
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Secondary Disabilities associated with FASDs

• Mental health problems—94%
• Trouble with the law—83%
• Sexual misconduct—49%
• Disrupted school experiences—61%
• Problems with alcohol and/or drug use
• Dependent living and employment for life
Spectrum of Clinical Symptoms

• Intellectual disability
• ADD, ADHD
• Poor memory and recall

• Poor compliance
• Poor planning and impulsivity
• Abstraction difficulties
Severity

Severity depends on alcohol:

- Dosage
- Pattern of exposure
- Timing of exposure
- Duration of exposure
Treatment across the Life Span

- Unique challenges in each age
- Family education—convey message of hope and critical need for support
- Increased supervision throughout adolescence and early adulthood
- Proactive preparation for adulthood
- Plan for supported living and employment
  - Few individuals with FASDs achieve independence as adults
- Proactive mental health services
What do we need to do to educate patients and professionals about alcohol use in pregnancy?
Myths about Alcohol and FAS

• Less than 1 drink per day in pregnancy is OK.
• Drinking late in pregnancy is OK and makes labor easier.
• Beer and wine are not alcohol, and thus are not a problem.
• If I drank and have one child without FAS, I can drink and I won’t have another child with FAS.
• If FAS doesn’t run in my family, my child won’t get FAS.
• FAS is curable if diagnosed early.
Messages in Lay Patient Education or Media Materials

- Books and websites on pregnancy do not deal with the issue or state there is little danger from occasional social drinking during pregnancy (e.g., www.yourbabytoday.com).
- Most messages are pro-alcohol use or don’t mention the dangers of drinking during pregnancy.
In 2002, only 25% of OB/GYN texts address alcohol use in pregnancy and some of those suggest that some amount of alcohol is ok!

Surveys of OB/GYN, FP, and Pediatricians suggest that about 20% of those physicians feel that drinking up to 1 drink per day in pregnancy is safe.
Is it any wonder women are confused about what to do regarding their drinking behavior during pregnancy?
Government and Policy Response

- 1973: term “FAS” coined in US
- 1981: First Surgeon General’s warning about the harmful effects of alcohol in pregnancy
- 1996: Institute of Medicine Report on the harmful effects of alcohol during pregnancy
- 2002: Congress mandates CDC to develop diagnostic criteria for FAS
Government and Policy Response

- 2004: CDC releases their report on FAS diagnostic criteria and recommendation on prevention
  - All children screened for FAS
  - All women screened for alcohol use
- 2005: Second Surgeon General’s advisory on Alcohol use and Pregnancy
  - In addition to pregnant women, women considering or at risk for pregnancy should abstain from alcohol
..."There are many individuals exposed to prenatal alcohol who, while not exhibiting all of the characteristic features of FAS, do manifest lifelong neurocognitive and behavioral problems arising from this early alcohol exposure… It is estimated that for every child born with FAS, three additional children are born who may not have the physical characteristics of FAS but still experience neurobehavioral deficits resulting from prenatal alcohol exposure that affect learning and behavior.”
Alcohol Use in Pregnancy: Who is to Blame?

- Widespread misunderstanding
  - Among women as well as healthcare providers
  - In health literature
- Alcohol is highly addictive
  - Women need assistance, not blame
  - Education is just the beginning
- Media blurs prevention messages
  - Pairing of sex and drinking
  - Enthusiastic report of healthy benefits
Drinking and Risky Sexual Behavior

- Age of drinking onset influences unplanned sex
- Alcohol influences decisions about sex
  - 29% 15-17 endorse
  - 37% 18-24 endorse
- Had unprotected sex due to alcohol use
  - 12% 15-17 endorse
  - 25% 18-24 endorse
FASD Prevention Messages

• No safe time, no safe amount, no safe type of alcohol in pregnancy
• If you are pregnant, don’t drink
• If you are drinking, don’t get pregnant
• If you can’t quit, get help
• The effects of alcohol on the fetus are permanent
Ethical and Legal Issues
Legal and Policy Issues

- Fetal rights and the maternal-fetal relationship
- Limitations of coercive and punitive approaches
- A public health approach
Fetal Rights and the Maternal-Fetal Relationship

**Ethical perspective**
- Evaluated in terms of personhood
  - Does the fetus have human rights?
  - Ethical status of fetus not settled, but well-being of each person starts during fetal development

**Legal perspective**
- Federal law—fetuses are not given the legal status of “persons,” with rare exception
- State law—varies by state
- Well-being—woman and fetus affected by well-being of each other
Limitations of Coercive and Punitive Approaches

- Law enforcement measures
  - Wisconsin—pregnant women whose habitual drinking exposes a fetus to substantial risks of physical harm taken into custody for involuntary inpatient alcohol treatment

- Legal decisions
  - Whitner vs. State of South Carolina (1997)—only state that permits criminal prosecution of women for endangerment of fetus
  - Ferguson v. City of Charleston (2001)—limits how health care providers at public hospitals can intervene to prevent fetal alcohol exposure
Limitations of Coercive and Punitive Approaches

- Acknowledgement of contributing factors in maternal drinking
  - Alcohol and drug addiction are illnesses
  - Women don’t intend to harm themselves and their fetuses
  - Role of treatment programs for alcohol and drug abuse

- Threatening and incarcerating women do not produce a lasting reduction in risk exposure
Six Objections to Punitive and Coercive Legal Approaches to Maternal Decision Making

- Coercive and punitive legal approaches to pregnant women who refuse medical advice fail to recognize that all competent adults are entitled to informed consent and bodily integrity.

- Court-ordered interventions in cases of informed refusal, as well as punishment of pregnant women for their behavior that might put a fetus at risk, neglect the fact that medical knowledge and predictions of outcomes in obstetrics have limitations.

- Coercive and punitive policies treat medical problems such as addiction and psychiatric illness as if they were moral failings.
Six Objections to Punitive and Coercive Legal Approaches to Maternal Decision Making

- Coercive and punitive policies are potentially counterproductive in that they are likely to discourage prenatal care and successful treatment, adversely affect infant mortality rates, and undermine the physician-patient relationship.
- Coercive and punitive policies directed toward pregnant women unjustly single out the most vulnerable women.
- Coercive and punitive policies create the potential for criminalization of many types of otherwise legal maternal behavior.
Supporters of Non-punitive Approaches

- American College of Obstetricians and Gynecologists
- American Academy of Pediatrics
- American Medical Association
- American Nurses Association
- American Public Health Association
- National Council on Alcoholism and Drug Dependence
- March of Dimes
A Public Health Approach

- Education and prevention as an alternative to punitive interventions
  - “When you drink, your baby drinks”
  - Effective use of contraception
  - Preconception abstinence from alcohol
Signage Laws: An Example of Effective Public Policy

- Signs reach broad audiences
- Opportunities for projects
- MRFASTM states with signage laws:
  - Missouri, Nebraska, Illinois

*WARNING: DRINKING ALCOHOLIC BEVERAGES DURING PREGNANCY MAY CAUSE BIRTH DEFECTS.*
A Public Health Approach: Challenges

- Many women do not know the dangers of alcohol to a fetus
- Social norms tolerate or even encourage drinking in pregnancy
- Health care providers reluctant to address alcohol use with female patients
  - Lack of training
  - Discomfort
  - Lack of time during health visits
A Public Health Approach: Challenges

- Drug and alcohol abuse treatment needed for women with alcohol dependence and abuse
- Screening by health care providers
- Few treatment options available for pregnant women and women with dependent family members
As practitioners, you deal with the ethical principles of confidentiality and autonomy on a daily basis.

In working with families with children with FAS, many of the legal/ethical issues you will encounter relate to the conflict between maternal and child rights.
Case Example

- You are Mrs. G’s primary care provider.
- She is pregnant with her third child.
- Her second child was born with FAS and removed from the home.
- Mrs. G has confided in you that she is drinking.
She says:

“Please don’t tell my caseworker I’m drinking.”
Ethical Issues

What is your response?
Would you…

- Honor her request?
- Report her immediately?
- Ask her further questions to get more information?
- Do a brief intervention for her drinking behavior?
Your ethical responsibilities include both Mrs. G and her fetus.

Most physicians do not report mothers who drink during pregnancy.
Confidentiality in the patient-provider relationship is founded on several important elements:

- First, on their right to privacy.
- Secondly, on their trust in you as a necessary component of the therapeutic relationship.
Ethical Issues

- There are exceptions to confidentiality:
  - The patient is a danger to herself.
  - The patient is a danger to the community or a third party.
  - The patient gives consent.
Mrs. G’s dilemma is a difficult one, but there are some steps you can take to make a decision:

1. Gather information.
2. Consider your “gut reaction.”
3. Identify the ethical problems.
4. Know your clinical duties.
5. Develop alternative courses of action.
Ethical Decision Making

5. Keep principles in mind.
6. Know your legal and professional requirements.
7. Make a decision.
8. Justify your decision.
10. Implement and document.
Summary

- Prevention education is health provider’s first responsibility to prevent FASDs.
- Health care providers have ethical responsibility to facilitate help for women and her child.
- Ethical care well-informed laws and policies strive for universal prevention measures and benefits for those affected by FASDs.
For further information

• Check our website: www.mrfastc.org

• Training in our 7 core competencies is available
Resources

- www.fascenter.samhsa.gov
- www.depts.washington.edu/fadu
- www.niaaa.nih.gov
- www.cdc.gov/ncbddd/fas/
- www.nofas.org
- www.thearc.org
- www.mimh.edu.fas/
- www.fasday.com/
- www.fasiceberg.com/