

POCKET GUIDE FASD MIRJAM N. LANDGRAF & FLORIAN HEINEN

THINK KIDS

DON'T DRINK
STOP FASD Fetal Alcohol
Spectrum Disorder

SOCIETY FOR NEUROPEDIATRICS, SWITZERLAND, AUSTRIA AND GERMANY



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THE 4 DIAGNOSTIC COLUMNS OF FAS

POSSIBLE RISK FACTORS FOR THE DEVELOPMENT OF FASD

MATERNAL ALCOHOL AND DRUG USE

- High alcohol intake
- Chronic alcohol abuse
- Alcohol use during the 1st & 2nd trimester (in contrast to use only during the 3rd trimester)
- Alcohol consumption throughout the whole pregnancy
- Additional intake of amphetamines and other illicit drugs

MATERNAL RISK FACTORS

- Age > 30 years
- Specific ethnicities
- Low socioeconomic status
- Malnutrition, lack of micronutrients and vitamins
- Stress
- Perinatal complications
- Siblings with FASD
- Genetic background

For the diagnosis of FAS, **each** of the following four criteria should be fulfilled.

1

GROWTH DEFICITS

2

FACIAL ANOMALIES

3

CNS ABNORMALITIES

4

CONFIRMED OR UNCONFIRMED
INTRAUTERINE ALCOHOL EXPOSURE

If a child presented to the health and social care system shows abnormalities in any one of the four diagnostic fields, each of the other three diagnostic fields should also be assessed. This may require referral to an appropriately qualified expert.



DIFFERENTIAL DIAGNOSIS REGARDING FASD
IN CHILDREN AND ADOLESCENTS –
without claiming complete listing

1. GROWTH DEFICITS

1.1. PRENATAL GROWTH DEFICITS

1.1.1. NORMAL INTRAUTERINE SUPPLY (FETAL PATHOLOGY)

ENDOGENOUS

- Malformation
- Genetic disorder (e.g. Turner's syndrome, Silver-Russel syndrome)
- Metabolic disorder

EXOGENEOUS

- Intrauterine infection (e.g. rubella, cytomegaly, toxoplasmosis, Herpes simplex, HIV, EBV, Parvovirus B19)
- Radiation exposure

1.1.2. IMPAIRED INTRAUTERINE SUPPLY

PREPLACENTAL

MATERNAL CONDITIONS

- Preeclampsia, hypotension, anemia, cyanotic vitia, collagen-vascular diseases, renal diseases,
- toxic effects, nicotine, drugs
- High maternal psychosocial stress

PLACENTAL

- Placenta previa (uterine malformation, myomata)
- Defective placentation
- Focal chromosome disorder restricted to the placenta

1.2. POSTNATAL GROWTH DEFICITS

- Familial hyposomia
- Constitutional retardation
- Skeletal dysplasia (e.g. hypochondroplasia, achondroplasia, osteogenesis imperfecta)
- Metabolic disorders
- Renal diseases
- Hormonal disorders
- Genetic syndromes (e.g. trisomy 21)
- Chronic diseases
- Malabsorption or malnutrition (e.g. vitamin D, calcium, and protein deficiency, general hypoalimentionation)
- Psychosocial hyposomia

To fulfill the criterion

GROWTH DEFICITS

at least one of the following abnormalities, documented at any time, should be present when taking into account gestational age, age and gender.

(1) Birth weight or body weight \leq 10th percentile

(2) Birth length or body length \leq 10th percentile

(3) Body Mass Index \leq 10th percentile

Microcephaly see 3.2.



To fulfill the criterion

FACIAL ANOMALIES

all three facial anomalies should be present:

DIFFERENTIAL DIAGNOSIS
REGARDING FASD IN CHILDREN AND ADOLESCENTS –
without claiming complete listing

2. FACIAL CHARACTERISTICS

2.1. TOXIC EFFECTS DURING PREGNANCY

- Anticonvulsive drugs
- Toluol
- Maternal phenylketonuria

2.2. GENETIC DISEASES

- Aarskog syndrome
- Cornelia de Lange's syndrome
- Dubowitz' syndrome
- Noonan's syndrome
- Williams-Beuren syndrome
- DiGeorge's syndrome
- Blepharophimosis syndrome
- Hallermann-Streiff syndrome
- 3-M syndrome
- Smith-Lemli-Opitz syndrome
- SHORT syndrome
- Feingold syndrome
- Kabuki syndrome
- Peter's-Plus syndrome
- Rubinstein-Taybi syndrome
- Geleophysic dysplasia

(1) **Short palpebral fissure length**
($\leq 3^{\text{rd}}$ percentile)

(2) **Smooth philtrum**
(Rank IV or V
Lip-Philtrum-Guide)

(3) **Thin upper lip**
(Rank IV or V
Lip-Philtrum-Guide)



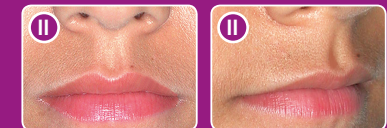
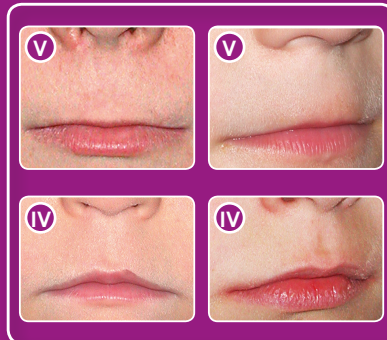
Measuring
palpebral fissure length



Measuring with
reference point



Measuring with a ruler



Lip-Philtrum-Guide

DIFFERENTIAL DIAGNOSIS REGARDING FASD
IN CHILDREN AND ADOLESCENTS –
without claiming complete listing

3. CNS ABNORMALITIES

3.1. FUNCTIONAL CNS DEFICITS

- Combined developmental disorders
- Intellectual deficits varying grades
- Developmental disorder of speech and language
- Developmental disorder of motor functions
- Attention deficit hyperactivity disorder
- Hyperkinetic disorder of social behavior
- Social behavior deficits with oppositional defiant behavior
- Combined social behavior and emotional deficits
- Stereotypy
- Aggressiveness
- Delinquency
- Disorders of addiction
- Childhood reactive attachment disorder
- Posttraumatic stress disorder
- Sexual deviation
- Sleep disorder
- Anxiety disorder /panic disorder
- Affective disorder
- Depressive disorder
- Epilepsies of other origin

3.2. MICROCEPHALY

- Familial microcephaly
- Genetic syndromes (see 2.2.)
- Prenatal malnutrition, toxic damage, infection
- Hypoxic ischemic cerebral damage
- Maternal diseases
- Postnatal malnutrition
- Metabolic disorders
- Chronic diseases

To fulfill the criterion

CNS ABNORMALITIES

3.1 or /and 3.2 should apply:

3.1

To meet the criterion
FUNCTIONAL CNS DEFICITS

at least one of the following abnormalities, that is not adequate for the patient's age and that cannot be explained solely by the familial background or social environment, should be present:

- (1) General intellectual deficits at least two standard deviations below the mean
or significant combined developmental delay in children under the age of two years
- (2) Performance at least two standard deviations below the mean in at least three of the following domains
or in at least two of the following domains combined with epilepsy:
 - Language /Speech
 - Fine motor functions
 - Spatial-visual perception or spatial-constructive skills
 - Learning or memory skills
 - Executive functions
 - Arithmetic skills
 - Attention
 - Social skills and behavior

3.2

To meet the criterion
STRUCTURAL CNS DEFICITS

The following deficit, documented at any time, **should** be present when taking into account gestational age, age and gender:

Microcephaly \leq 10th percentile / \leq 3rd percentile



CONFIRMED OR UNCONFIRMED INTRAUTERINE ALCOHOL EXPOSURE

POTENTIAL RISK FACTORS FOR MATERNAL ALCOHOL CONSUMPTION DURING PREGNANCY

AGE

- > 30 years
- binge drinking < 27 years

NATIONALITY

- No migration background
- High acculturation
- Specific minorities (e.g. Native Indians, Inuits)

HEALTH RELATED RISK FACTORS

- Starting to drinking alcohol early in life
- Alcohol consumption, especially binge drinking, before pregnancy
- Previously treated for alcohol-related health problems
- Illicit drug use
- Smoking

CHARACTERISTICS OF THE PREGNANCY

- Unplanned or unwanted pregnancy
- Substandard or late prenatal care

SOCIOECONOMIC STATUS

- High socioeconomic status
- Dependent on public support

SOCIAL ENVIRONMENT

- Single or unmarried
- Alcohol or drug use within the family or by the partner
- Low social support

PSYCHOLOGICAL FACTORS

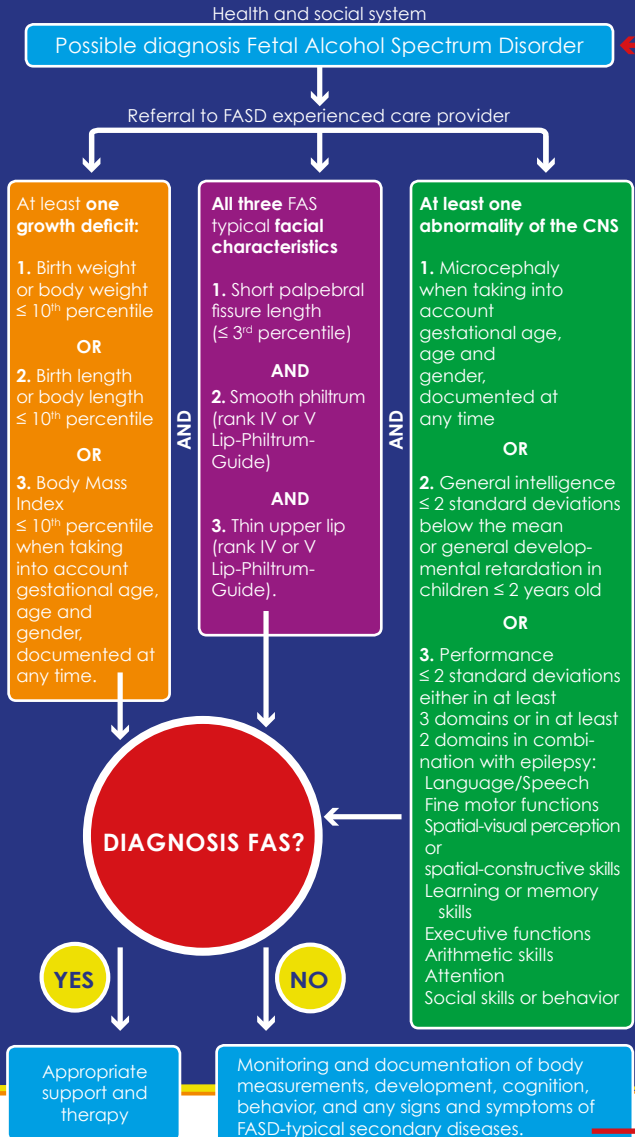
- Previous or current physical or sexual abuse by the partner or others
- Psychological and psychiatric disorders including depression, anxiety and panic disorder, sexual dysfunctions

Even if maternal alcohol consumption during pregnancy is not confirmed, the patient's condition **should** be diagnosed as FAS if the criteria of the three other diagnostic fields are met.



ALGORITHM

DIAGNOSTIC WORKUP FOR FETAL ALCOHOL SYNDROME



THE **3** DIAGNOSTIC COLUMNS OF PFAS

For the diagnosis of **pfAS** –
partial Fetal Alcohol Syndrome –
all 3 criteria below should be fulfilled:

- 1** FACIAL ANOMALIES
- 2** CNS ABNORMALITIES
- 3** CONFIRMED OR PROBABLE
INTRAUTERINE ALCOHOL EXPOSURE



To fulfill the criterion

FACIAL ANOMALIES

2 of 3 facial anomalies should be present
(documented at any time):

(1) Short palpebral fissure length
($\leq 3^{\text{rd}}$ percentile)

(2) Smooth philtrum
(Rank IV or V
Lip-Philtrum-Guide)

(3) Thin upper lip
(Rank IV or V
Lip-Philtrum-Guide)



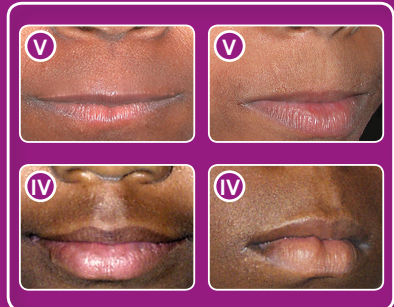
Measuring palpebral fissure length



Measuring with reference point



Measuring with a ruler



Lip-Philtrum-Guide

To fulfill the criterion

CNS ABNORMALITIES

Persons of the professional-supportive or private environment, who are able to give reliable information about maternal alcohol consumption during pregnancy, **should** be asked when taking the medical history from a third party. Regulatory and legal framework for the exchange and transfer of information **should** be considered (expert consensus).

at least 3 of the following abnormalities, that are not adequate for the patient's age and that cannot be explained solely by the familial background or social environment, should be present:

- General intellectual deficits at least two standard deviations below the mean **or** significant combined developmental delay in children under the age of two years
- Epilepsy
- Microcephaly

Performance at least 2 standard deviations below the mean:

- Language / Speech
- Fine motor functions and coordination
- Spatial-visual perception or spatial-constructive skills
- Learning or memory skills
- Executive functions
- Arithmetic skills
- Attention
- Social skills and behavior



CONFIRMED OR PROBABLE INTRAUTERINE ALCOHOL EXPOSURE

The guideline group **defines** “probable maternal alcohol consumption during pregnancy” as oral or written information in the medical history from a third party.

Persons from the private environment could be (respecting any possible family conflicts):

- Father of the child
- Partner living with the mother during pregnancy
- Other relatives of the child
- Other caregivers, who are in close contact with the mother during pregnancy

Persons of the professional-supportive environment can be:

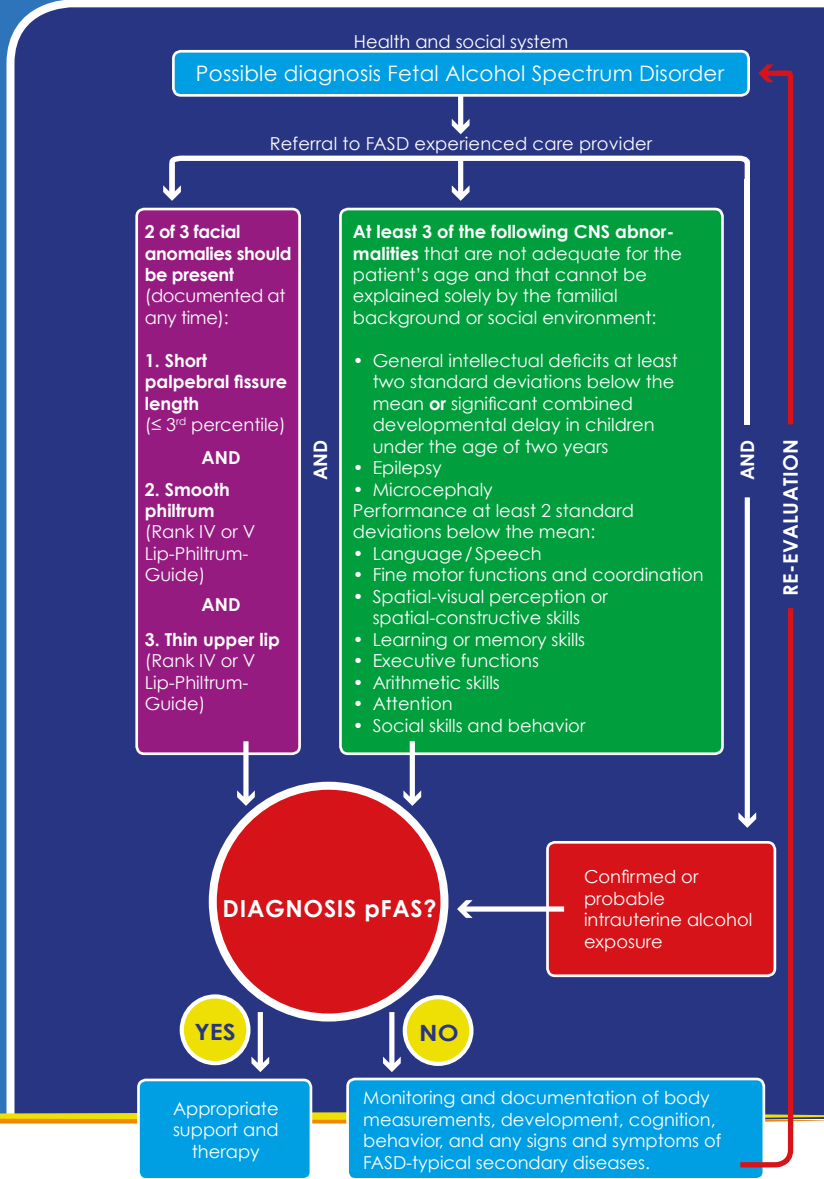
- Midwives
- Family doctors
- Youth welfare service agents

If the maternal alcohol consumption during pregnancy is **probable or confirmed** and the patient has facial anomalies and CNS abnormalities, the patient's condition should be diagnosed as pFAS.



ALGORITHM

DIAGNOSTIC WORKUP FOR PARTIAL FETAL ALCOHOL SYNDROME



THE **2** DIAGNOSTIC COLUMNS OF ARND

ARND is a "not visible disability" of the child / adolescent. The diagnosis is difficult and can only be attained by a complex psychological assessment.

For the diagnosis of **ARND** – alcohol related neurodevelopmental disorder – **the 2 criteria below should** be fulfilled:

1 CNS ABNORMALITIES

2 CONFIRMED INTRAUTERINE ALCOHOL EXPOSURE



To fulfill the criterion

CNS ABNORMALITIES

Recommendation:

Because the diagnosis of ARND is complex and in its differentiation to other developmental disorders difficult, the guideline group recommends referring the child / adolescent with suspected ARND to a care provider, experienced in FASD (expert consensus).

at least 3 of the following abnormalities, that are not adequate for the patient's age and that cannot be explained solely by the familial background or social environment, should be present:

- General intellectual deficits at least two standard deviations below the mean **or** significant combined developmental delay in children under the age of two years
- Epilepsy
- Microcephaly

Performance at least 2 standard deviations below the mean:

- Language / Speech
- Fine motor functions and coordination
- Spatial-visual perception or spatial-constructive skills
- Learning or memory skills
- Executive functions
- Arithmetic skills
- Attention
- Social skills and behavior



Confirmed INTRAUTERINE ALCOHOL EXPOSURE

Currently, the significance of the amount of maternal alcohol consumption during pregnancy can only be estimated qualitatively because a reliable cut-off for a harmless intrauterine alcohol exposure for the unborn child does not exist.

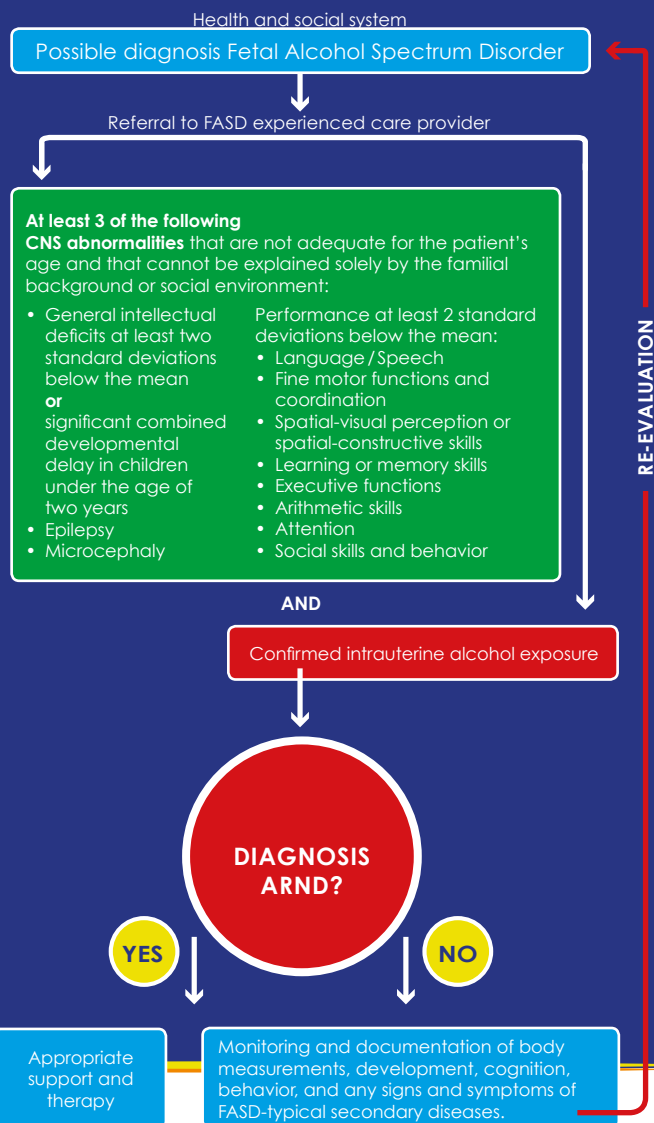
Based on international studies, a repetitive intake of alcohol or at least one binge drinking episode (≥ 5 drinks per occasion) during the pregnancy poses the child at risk to develop FASD.

If the maternal alcohol consumption during pregnancy is **confirmed** and the patient has CNS abnormalities, the patient's condition should be diagnosed as ARND.



ALGORITHM

DIAGNOSTIC WORKUP FOR ALCOHOL RELATED NEURODEVELOPMENTAL DISORDER



! ARBD: ALCOHOL RELATED BIRTH DEFECTS

Alcohol related birth defects (ARBBD) **should** not be used as a diagnostic term because of the lack of specificity of the malformations and the lack of evidence for ARBD as separate entity of disease.



**Guideline Diagnosis of
Fetal Alcohol Spectrum Disorders
(Germany)**

Short version, long version and methodological report
<http://www.awmf.org/leitlinien/detail/ll/022-025.html>

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**Homepage of the Addiction-Authorized
Representative of the German Government**

www.drogenbeauftragte.de

Federal Center for Health Education (Germany)

www.bzga.de