Use of Epic “SmartForms” for Diagnosis in the FASD Clinic

Fetal Alcohol Spectrum Disorders (FASD)is an umbrella term which encompasses a group of similar, but non-overlapping conditions: Fetal Alcohol Syndrome (FAS), Partial Fetal Alcohol Syndrome (pFAS), Alcohol Related Neurobehavioral Disorder (ARND), Alcohol Related Birth Defect (ARBD). Neurobehavioral disorder associated with prenatal alcohol exposure (ND-PAE) is a psychiatric condition described in the DSM-V which is often included in FASD. FASD was initially described in 1996, and in 2005, clinical guidelines regarding diagnostic criteria of each condition was proposed. Updates to the diagnostic criteria of FASD were published in 2016 (Hoyme, et al., 2016).

The goal of this project was to develop a specific tool within the institution’s (Rady Children’s Hospital) electronic medical record (EMR) to facilitate diagnosis of FASD. A secondary goal was to create a tool capable of mining specific data points about the patients of the FASD clinic, including physical exam findings, congenital malformations, and neuropsychological profiles. This will be useful in future research studies conducted with the FASD team. This project was conducted with Dr. Miguel Del Campo, with input from various members of the FASD clinic, and in close collaboration with the Rady IT department.

Rady Children’s Hospital utilizes the EPIC EMR in both its clinics and inpatient facilities. It is a customizable EMR. After first outlining the format of the various questionnaires which would be used to gather the appropriate data to establish a diagnosis of FASD, the outlines were then converted into EPIC SmartForms. SmartForms allow data to be encoded and captured while concurrently writing a clinic visit or progress note. With the help of the FASD clinic providers, the SmartForms were edited to fit within the existing clinic workflow. Furthermore, the SmartForms may be edited in response to clinician feedback and with evolving diagnostic guidelines.

The first SmartForm records prenatal exposure history. This is adapted from Table 2: Definition of Documented Prenatal Alcohol Exposure of the *“*Updated Clinical Guidelines for Diagnosing Fetal

Alcohol Spectrum Disorders". A positive response to any of the six questions is interpreted by the EMR as confirmed prenatal alcohol exposure. A seventh question asks if the biological mother or close family member specifically denies prenatal alcohol exposure. When this question is answered positively, and either the mother or family member denies PAE, the EMR interprets this as no prenatal alcohol exposure. In the cases where the biological mother is unavailable, or declines to answer, there EMR will record prenatal alcohol exposure as "unknown".

In addition to the prenatal alcohol exposure history, this SmartForm also captures the following as discrete data: the date of recognition of pregnancy, the amount of drinks per month of pregnancy, other possible drug prenatal exposures, and the identity of the reporting party and their relationship to the child.

1. Prenatal Alcohol Exposure SmartForm

	Questions	Yes	No	Unavailable
1.	≥6 drinks/week for ≥2 weeks during pregrancy			
2.	≥3 drinks per occasion on ≥2 occasions during pregnancy			
3.	Documentation of alcohol-related social or legal problems in proximity to (before or during) index pregnancy			
4.	Documentation of intoxication during pregnancy by blood, breath, or urine alcohol content testing			
5.	Positive testing with established alcohol-exposure biomarkers during pregnancy or at birth			
6.	Increased prenatal risk associated with drinking during pregnancy as assessed by a validated screening tool			
7.	Does the biological mother or other close family member specifically deny prenatal alcohol exposure?			

A second SmartForm is used to record the minor facial anomalies characteristic of FASD which include short palpebral fissures (less than 10th percentile), thin vermillion border, and smooth philtrum (ranked 4-5 on lip/philtrum guide). At least 2 of these features must be present to make the diagnosis of FAS or pFAS. A height or weight deficiency (less than 10th percentile in either) is also required to make a diagnosis of FAS or pFAS without confirmed alcohol exposure. Ideally, these values will be pulled

in to the SmartForm automatically with the MA or nurse is rooming the patient and taking vitals.

Decreased head circumference (less than 10th percentile) is a marker for abnormal brain development.

2. Minor Facial Anomalies SmartForm

Minor Facial Anomalies	Measurement		Percentile ≤10%ile		
Right palpebral fissure			Yes		No
Left palpebral fissure			Yes		No
Philtrum	**1**	**2**	**3**	**4**	**5**
Vermillion border	**1**	**2**	**3**	**4**	**5**

Growth	Measurement	Percentile (calculated)
Height (3)		
Weight (3)		

Though not specifically required for the diagnosis of any of the conditions included in FASD, children with prenatal alcohol exposure tend to have certain physical findings in addition to the minor facial anomalies listed above. These include abnormal hand creases, joint contractures, as well as many others. In this SmartForm, a list of known physical findings is found grouped according to body part. If present in the examined child, a plus sign may be checked, if absent, a minus sign may be checked.

3. Minor Physical Features of FASD SmartForm

Head/Face	
Hypoplastic midface (2)	
Prognathism (1)	
Other	

Ears		
Railroad track ears (1)	R	L
Other	R	L

Eyes		
Strabismus (1)	R	L
Ptosis (1)	R	L
Epicanthal folds (2)	R	L
Other	R	L

Nose	
Flat nasal bridge (2)	
Anteverted nares (2)	
Other	

Hands		
Hockey stick crease (2)	R	L
Single transverse crease (2)	R	L
Hypoplastic thenar crease (2)	R	L
Other aberrant crease (2)	R	L
Camptodactyly (2)	R	L
Clinodactyly of 5th finger (2)	R	L
Hypoplastic nails (1)	R	L
Other	R	L

Arms		
Decreased pronation/supination of elbow (1)	R	L
Other	R	L

Hips/Legs/feet		
Knee contractures	R	L
Hip contractures	R	L
Other contractures	R	L
Other	R	L

Other Body	
Scoliosis	
Pectus carinatum	
Pectus excavatum	
Hypertrichosis (1)	
Other	

Heart	
Murmur (1)	
Other	

Mental status/Behavior	
Hyperactive	
Other	

Neurological	
Hypertonic	
Hypotonic	
Seizures	
Other	

The following SmartForm records the presence or absence of major congenital malformations. The diagnosis of ARBD requires the presence of a major malformation that has been reported with increased frequency in human newborns prenatally exposed to alcohol and/or has been shown in animal studies to be the consequence of prenatal alcohol exposure. When prenatal alcohol exposure is present, and one of the qualifying malformations is marked by the provider, the EMR will automatically calculate a diagnosis of ARBD.

4. Major Malformations SmartForm

Cardiac	
ASD	
VSD	
Aberrant great vessels	
Conotruncal heart defects	
Other	

Skeletal	
Radioulnar synostosis	
Vertebral segmentation defects	
Large joint contractures	
Scoliosis	
Other	

Renal	
Aplastic kidney	
Hypoplastic kidney	
Dysplastic kidney	
Horseshoe kidney	
Ureteral duplications	
Other	

Eyes	
Retinal vascular anomalies	
Optic nerve hypoplasia	
Refraction error	
Strabismus	
Ptosis	
Other	

Ears	
Conductive hearing loss	
Neurosensory hearing loss	
Other	

The updated guidelines included structural brain anomalies and recurrent non-febrile seizures as possible ways in which to meet the criteria for deficient brain growth, in addition to a small head circumference. Published in the European Journal of Medical Genetics, the article "Correlation Between Morphological MRI Findings and Specific Diagnostic Categories in Fetal Alcohol Spectrum Disorders", provides a starting point for the structural brain anomalies consistent with a diagnosis of FASD. If a brain MRI is available for the patient, this SmartForm will list the possible findings which meet the criteria for deficient brain growth. Data for history of non-febrile seizures and EEG findings if available will also be collected.

5. Brain Abnormalities SmartForm

Brain MRI	
Corpus callosum hypoplasia	
Cerebellar malformations	
Vascular anomalies	
Focal gliosis	
Dilated perivascular spaces	
Cervical vertebral segmentation anomalies	
Occipitocervical junction anomalies	
Pituitary hypoplasia	
Ventriculomegaly	
Arachnoid cysts	

Cavum septum pellucidum	
Simplified gyral pattern	
Optic chiasma hypoplasia	
Incomplete hippocampal inversion	
Fornix dysplasia	
Periventricular heterotopias	
Cortical malformations	
Other	

History of recurrent non-febrile seizures?	Yes	No

EEG Findings	
Abnormal background	
Generalized IEDs	
Generalized non-specific paroxysms	
Focal IEDs	
Focal non-epileptiform discharges	
Epilepsy	
Febrile seizures	

This next SmartForm captures data regarding neuropsychological testing that the patient may have undergone. If available, these test results are useful in making the diagnosis of ND-PAE. The profile is divided into three categories: neurocognitive impairment, impairment in self-regulation, and deficits in adaptive functioning. While neurocognitive impairment or impairment in self-regulation is sufficient to meet the criteria for neurobehavioral impairment in FAS or pFAS, ND-PAE requires impairment in each category.

6. Neurobehavioral SmartForm

1. Neurocognitive impairment	
Global intellectual impairment	
Impairment in executive functioning	
Impairment in learning	
Impairment in memory	
Impairment in visual special reasoning	

2. Impairment in self-regulation	
Impairment in mood/behavioral regulation	
Attention deficit	
Impairment in impulse control	

3. Deficits in adaptive functioning	
Communication deficit	
Social communication and interaction impairment	
Impairment in daily living	
Motor impairment	

Using the data gathered from the SmartForms, the EMR will be able to determine which patients meet the criteria for a diagnosis on the conditions in FASD. The following table demonstrates how the diagnosis is calculated.

7. Diagnosis Algorithm

Diagnosis	Criteria	Assessment Statement
Prenatal alcohol exposure ICD-10: P04.3	Prenatal Alcohol Exposure (Section I)	*Child name* meets the criteria for prenatal alcohol exposure.
Fetal alcohol syndrome with confirmed alcohol exposure ICD-10: Q86.0	≥2 Minor Facial Anomalies (Section IIa) AND Growth deficiency (Section IIb) AND Brain abnormality (Section II c, OR Section V a, OR Section V b) AND Neurobehavioral impairment (Section IV a)	*Child name* meets the criteria for a diagnosis of fetal alcohol syndrome based on the following criteria: a characteristic pattern of minor facial anomalies, growth deficiency, brain abnormalities, and neurobehavioral impairment.
Partial fetal alcohol syndrome ICD-10: Q86.0	Prenatal Alcohol Exposure (Section I) AND ≥2 Minor Facial Anomalies (Section IIa) AND Neurobehavioral impairment OR ≥2 Minor Facial Anomalies (Section IIa)	*Child name* meets the criteria for a diagnosis of partial fetal alcohol syndrome based on the following criteria: confirmed prenatal alcohol exposure, a characteristic pattern of minor facial anomalies, and neurobehavioral impairment. OR *Child name* meets the criteria

	AND Neurobehavioral impairment (Section IV a) AND EITHER Growth deficiency (Section IIb) OR Brain abnormality	for a diagnosis of partial fetal alcohol syndrome based on the following criteria: a characteristic pattern of minor facial anomalies, neurobehavioral impairment and either growth deficiency or brain abnormality.
Alcohol related neurobehavioral disorder (ARND) ICD-10: Q86.0	Prenatal Alcohol Exposure (Section I) AND Neurobehavioral impairment (Section IV a)	*Child name* meets the criteria for a diagnosis of ARND based on the following criteria: confirmed prenatal alcohol exposure and neurobehavioral impairment.
Alcohol related birth defect (ARBD) ICD-10: Q86.0	Prenatal Alcohol Exposure (Section I) AND ≥1 Major Malformation (Section IV)	*Child name* meets criteria for a diagnosis of ARBD based on the following criteria: confirmed prenatal alcohol exposure and at least one major malformation resulting from that exposure.
Neurobehavioral disorder associated with prenatal alcohol exposure ICD-10: Q86.0	Prenatal Alcohol exposure (Section I) AND Neurocognitive impairment (Section IV) AND Impairment in self-regulation (Section IV) AND Deficits in adaptive functioning (Section IV)	*Child name* meets criteria for a diagnosis of ND-PAE based on the following: neurocognitive impairment, impairment of self-regulation, and deficits in adaptive functioning in the context of known prenatal alcohol exposure.

In addition to streamlining diagnosis and capturing information for research, the integration of SmartForms into the clinical workflow may also be used to manage resources and referrals. While still a work in progress, this SmartForm will ideally identify the referring party to the FASD clinic, as well as track which services (Neuropsychological testing, KIDSTART, FASD support group etc.) are utilized by the patients. This will allow providers at the clinic to better meet the needs of patients affected by FASD.

References

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Hoyme, H. E., Kalberg, W. O., Elliot, A. J., Blankenship, J., Buckley, D., Marais, A.-S., . . . Jones, K. (2016). Updated Clinical Guidelines for Diagnosing Fetal Alcohol Spectrum Disorders. *Pediatrics*.