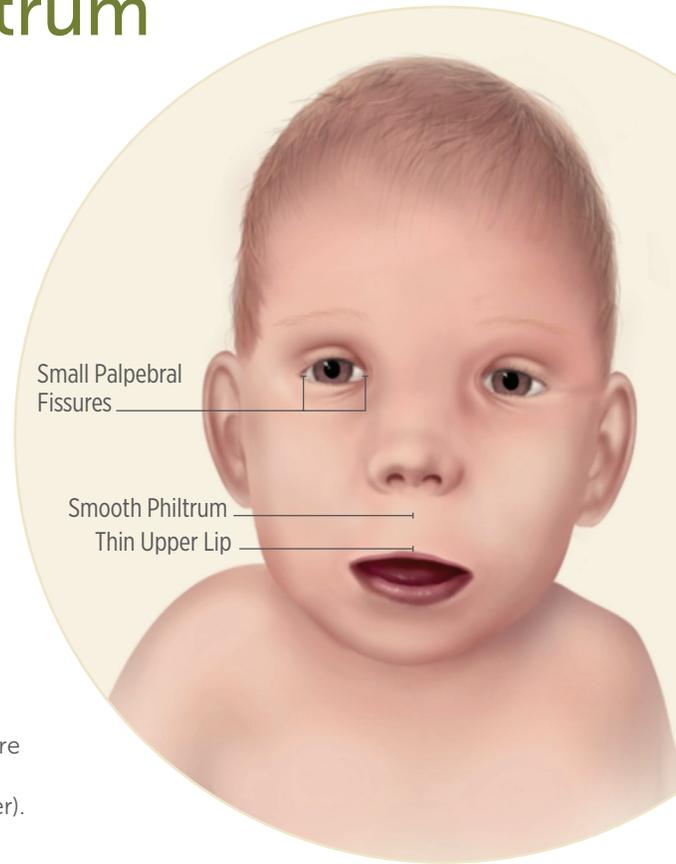


Dysmorphic Facial Features of Fetal Alcohol Spectrum Disorders (FASDs)

Minor abnormalities (dysmorphia) of facial features are very important clues to brain structure and function. They provide an outward sign on brain formation and development in gestation. Teratogens, such as alcohol, as well as genetic errors that disrupt brain development can have a downstream effect on formation of the skull and the coming together of midline facial features. Knowledge of features associated with certain diagnoses can be one piece that helps the pediatrician put together the puzzle for children with developmental, cognitive and/or behavioral problems.

Fetal alcohol spectrum disorders (FASDs) is an umbrella term that describes several conditions resulting from prenatal exposure to alcohol. Two of these diagnoses, Fetal Alcohol Syndrome (FAS) and Partial Fetal Alcohol Syndrome (pFAS), have associated dysmorphic features. The following is a brief description of the three sentinel or cardinal facial dysmorphic features relevant to these diagnoses. Dysmorphology, however, covers only 20% of the individuals with FASDs. This overview is intended to raise awareness of dysmorphic features in primary care pediatric medical home rather than teach diagnostic technique (which is best learned in 1:1 settings with an experienced examiner).



■ SMALL PALPEBRAL FISSURES (≤ 10th Centile)

Palpebral fissures are the horizontal opening of the eye. The measurement is from the endocanthion to the exocanthion for each eye in millimeters (mm). These measurements are converted to percentiles using norm-based charts (see example on reverse side). Clinically, small palpebral fissures will give the appearance of very wide-spaced eyes.

■ SMOOTH PHILTRUM

The philtrum is the groove that runs from the center root of the septum to the border of the upper lip. This feature varies on a continuum and assessment can be subjective. Thus, scales that rank the smoothness of the philtrum from 1 to 5 have been developed and validated in multiple sites (see lip/philtrum guide on reverse side). Separate scales are available for Caucasian and African American children.

■ FLATTENED OR THINNED UPPER VERMILION BORDER

The upper lip or border of the lip is thinned or non-existent. Again, because of the continuous nature and subjective nature of this measurement, a 1 to 5 ranking has been developed for the lip/philtrum guide.

Additional features that may be present, but not necessary for diagnosis:

Epicanthal Folds | 5th Finger Clinodactyly | Hockeystick Sign | Hypoplastic Nails | Camptodactyly | Shortened 5th Finger
Midface Hypoplasia | Railroad Track Sign | Difficulty with Pronation/Supination | Palate Abnormalities/Bifid Uvula

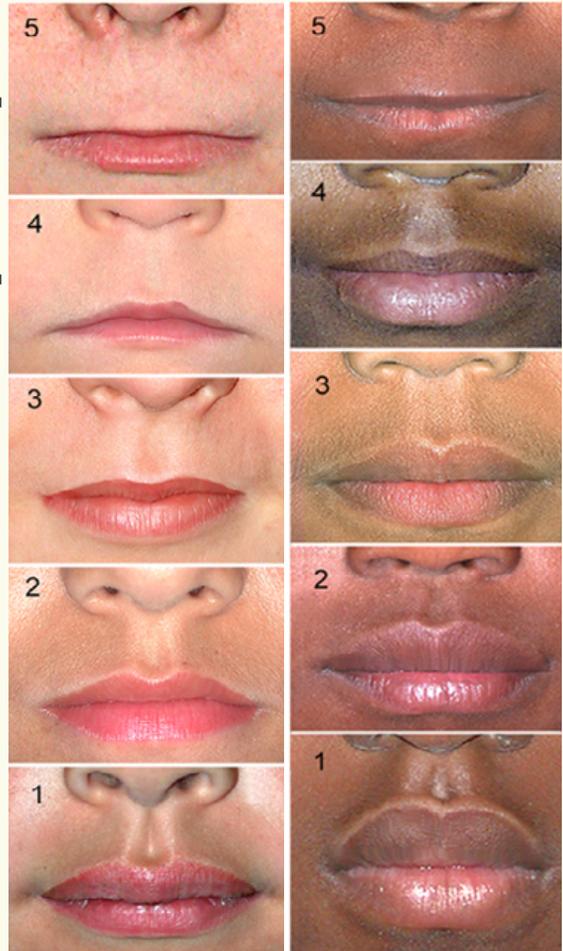
See additional information on reverse side.

Lip and Philtrum Guide

These photographs represent the range of smoothness and thinness that can occur for the philtrum and upper lip in Caucasian (left) and African American (right) children. Photos labeled 1, 2 and 3 show lip and philtrum within normal limits (number 3 is the general population mean). Photos labeled 4 and 5 show lip thinness and smooth philtrums that indicate prenatal exposure to alcohol and would meet criteria for an FAS/pFAS diagnosis.

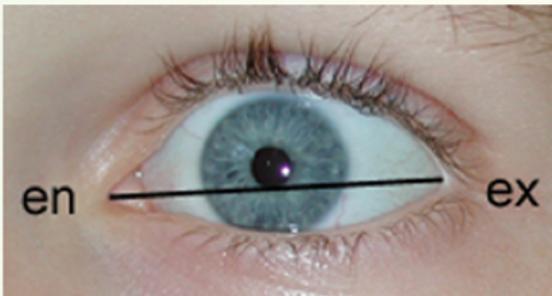
Photos copyright 2018, Susan Astley PhD, University of Washington. Reprinted by permission.

FAS



Palpebral Fissure Length Measurement: Endocanthion to Exocanthion

The image below shows a palpebral fissure length measurement. On a neutral, relaxed face, the measurement reflects the distance from the endocanthion to the exocanthion passing just under the pupil.



Palpebral Fissure Length Centiles Nomogram

Pictured on right is a nomogram for determining palpebral fissure length centiles.

Reprinted from Thomas, I.T., Gaitantzis, Y.A., Frias, J.L., 1987. Palpebral fissure length from 29 weeks gestation to 14 years. *Journal of Pediatrics* 111, 267-268.

