Section C: Assessing Sentinel Facial Features

Fetal exposure to alcohol during the first trimester affects development of facial features. The areas most affected are the orbital region (eyes) and mid-face. The effect of prenatal alcohol exposure on fetal brain growth is also thought to affect the size and shape of the face. A range of facial anomalies can occur as a result of prenatal alcohol exposure.

There are three features which commonly occur across age, gender and ethnic groups:

- **Small palpebral fissures**: Short horizontal length of the eye opening, defined as the distance from the endocanthion to the exocanthion (points A and B on photo below)
- **Smooth philtrum**: Diminished or absent ridges between the upper lip and nose
- **Thin upper lip**: With small volume

These features are shown in the photo below.

![Photo reproduced with permission from Susan Astley, University of Washington](image)

Although these facial features may also occur independently as normal variations in the general population (unrelated to prenatal alcohol exposure), when seen in combination, these facial features are pathognomonic of and highly specific to prenatal alcohol exposure. They are termed the ‘sentinel’ facial features of FASD.
Facial anomalies are one of the three diagnostic criteria for FASD, together with prenatal alcohol exposure and neurodevelopmental impairment. A diagnosis of FASD may be made with or without facial features.

- A diagnosis of **FASD with three sentinel facial features** means that the individual has all 3 of the characteristic (or ‘sentinel’) facial features that have been associated with prenatal alcohol exposure.

- A diagnosis of **FASD with less than 3 sentinel facial features** means that the individual may have 0, 1 or 2 of the characteristic facial features.

The University of Washington FAS Prevention and Diagnostic Network has developed criteria for FASD sentinel facial features:

- **Short palpebral fissure length** (PFL) 2 or more standard deviations below the population mean (or <3rd percentile). This equates to a z-score of -2 or more.
- **Smooth philtrum** – Rank 4 or 5 on the University of Washington Lip-Philtrum Guide
- **Thin upper lip** – Rank 4 or 5 on the University of Washington Lip-Philtrum Guide

Assessment can be using direct measurement and clinical examination and/or computerised analysis of a digital facial photograph (as described by Astley and Clarren (80, 81). Facial features may alter with age. Diagnosis should be based on the point in time when the features were most clearly expressed.

Further details regarding how to assess sentinel facial features are found in Appendix C.

**Considerations regarding assessment of sentinel facial features**

**Palpebral fissure length (PFL)**

PFL growth charts have been developed for populations overseas. In the absence of Australian reference data, we recommend using:

- **Scandinavian (Stromland) charts** if a child is under 6 years of age
- **Canadian (Clarren) charts** if a child, adolescent or adult is over 6 years

The Canadian charts are based on a multi-racial population considered to be a better representation of Australian children, although this has not been qualified by research. As the charts start at 6 years of age, Scandinavian charts need to be used in children under 6 years of age.

For infants and children under 2 years of age, the **corrected age of an ex-premature** child should be used if they are under 2 years of age (similar to other growth parameters such as head circumference, height and weight).

For older adolescent and adults, since PFL matures by 16 years without further changes, PFL norms and z scores for 16 year olds can be used for individuals over 16 years of age (from the Clarren charts).
Upper Lip Thinness and Philtrum Smoothness

Upper lip thinness and philtrum smoothness should be assessed using the University of Washington (UW) Lip-Philtrum Guides, which comprise photographs according to a 5 rank scale, which the range of lip thickness and philtrum depth seen in a population (i.e. the normal distribution).

- **Ranks 1, 2 and 3** are not associated with prenatal alcohol exposure, and are **below diagnostic threshold for FASD**
- **Ranks 4 and 5** are also caused by and characteristic of prenatal alcohol exposure and FASD but are also seen in a small proportion of the general population.

The University of Washington has developed guides for two ethnic populations: Caucasian (Guide 1) and African American (Guide 2) – see Appendix C. They recommend:

- Lip-Philtrum Guide 1 should be used for Caucasians and all races (or combinations of races) with lips like Caucasians.
- Lip-Philtrum Guide 2 should be used for African Americans and all races (or combinations of races) with thicker lips like African Americans.
Guides specific to Australian populations have not yet been developed, although research has commenced. In the absence of Australian lip-philtrum guides, the clinician should use charts which best fit the lip thickness of the individual they are assessing, while also considering the ethnic background/s of the individual.

Nonetheless, Lip-Philtrum Guides specific to every racial group may not to be required due to the lack of a homogenous phenotype for many races, the frequency of multiracial ancestry, and the small magnitude of differences involved.(18) In addition, small palpebral fissure length is the most consistent finding in 2D and 3D studies of facial features of FASD in different ethnic populations and ages, suggesting it is particularly sensitive to prenatal alcohol exposure. Smooth philtrum and thin upper lip are also consistent findings across populations. Recent studies indicate there are racial differences in other PAE related facial features (82, 83).

Other dysmorphic features
Other dysmorphic features have been observed in FASD but are not specific to FASD. These should be documented during assessment and include:

- **Facial features:** Flat nasal bridge, midface hypoplasia (flat midface), epicanthic folds, differences in craniofacial width, ear length and facial depth, widened intercanthal distance, anteverted nares (short upturned nose), micrognathia. (47, 84)

- **Other minor congenital anomalies:** clinodactyly (abnormal curving of the fifth finger toward the fourth finger), "Hockey stick" configuration of the upper palmar crease, other palmar crease abnormalities, “railroad track” ears, ptosis, strabismus, decreased elbow pronation/supination, incomplete extension of one or more digits, camptodactyly (permanent flexion of one or both finger interphalangeal joints, most commonly fifth and fourth fingers), shortened fifth digits. (47)

- Major birth defects of the cardiac, renal, ocular, auditory and skeletal systems such as optic nerve hypoplasia and septal defects. (4, 85, 86)

Individual dysmorphic features can occur in multiple syndromes and examination for features that differentiate alternate or co-existing syndromes and other disorders during the diagnostic assessment is essential. Differential diagnosis should include consideration of conditions that have a clinical presentation that is similar to FASD.(85)

If a genetic disorder is suspected, or any uncertainty regarding differential diagnosis exists, review by a clinical geneticist is indicated.

See Appendix D for Syndromes with constellations of features which overlap with FASD. (4)