Elements of Morphology: Standard Terminology for the Periorbital Region

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An international group of clinicians working in the field of dysmorphology has initiated the standardization of terms used to describe human morphology. The goals are to standardize these terms and reach consensus regarding their definitions. In this way, we will increase the utility of descriptions of the human phenotype and facilitate reliable comparisons of findings among patients. Discussions with other workers in dysmorphology and related fields, such as developmental biology and molecular genetics, will become more precise. Here we introduce the anatomy of the periorbital area and define and illustrate the terms that describe the major characteristics of the periorbital area.

Key words: terminology; definitions; periorbital structures; eyebrows; eyelashes; eyelids; palpebrae; telecanthus; lacrimal glands

INTRODUCTION

General

This article is part of a series of six articles defining the morphology of regions of the human body [Allanson et al., in press-b; Biesecker et al., in press; Carey et al., in press; Hennekam et al., in press; Hunter et al., in press]. The series is accompanied by an introductory article describing general aspects of this project [Allanson et al., in press-a]. The reader is encouraged to consult the introduction when using the definitions.

Anatomy

The general anatomy of the non-globe periorbital region is depicted in Figure 1. The definitions for the terms utilized in describing the features within this region are listed alphabetically. The anatomy of the various structures is described in more detail below.

Brow: The soft tissue at the junction of the frontalis and orbicularis oculi muscles, overlying the bony supraorbital ridge.

Eyebrow: The arch of hair on the brow (Fig. 2) [Goss, 1959]. The eyebrows usually extend further laterally than medially, in relation to the eye, and are wider and thicker medially. Based on observed localized abnormalities of the eyebrow, it is useful to divide the eyebrow into three parts: medial, middle (central), and lateral. The hairs of the medial part are oriented laterally, while those of the middle (central) part are oriented superolaterally. The transition between the middle and lateral parts is less frequently visible. Some syndromes have unique patterns of aberrations in one or more of these three areas. The eyebrow is sometimes referred to as the supercilium.

Eye spacing: There is wide variation in interorbital distance and in the placement of the canthi [Cohen et al., 1995]. A number of terms in this article address the nomenclature of these variations. Several of the terms are commonly confused (especially telecanthus and hypertelorism). Some of the variations are illustrated in Figure 3.

Eyelashes: Hairs that emanate from the margins of the eyelids [Goss, 1959].

Eyelid (syn. Blepharon, palpebra [plural: palpebrae]): A fold of skin and its subcutaneous components that covers the anterior globe. The upper lid is bounded by the soft tissue overlying the inferior border of the bony supraorbital ridge and inferiorly by the lid margin. The lower lid is bounded by the soft tissue overlying the infraorbital rim and superiorly by the lid margin.

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crescent-shaped crease on the upper eyelid represents the location of attachment of the levator palpebrae muscle to the orbicularis oculi muscle [Goss, 1959].

Lacrimal punctum [plural: puncta]: This structure represents the external aperture of the tear duct system. It can be absent, malpositioned, or obstructed [Ogawa and Gonnering, 1991], and several terms below address these findings.

Palpebral fissure: When the eye is open the palpebral fissure is a scaphoid space or outline formed by eyelid margins. The palpebral fissure extends from the lateral canthus (outer canthus) to the medial canthus (inner canthus). Many factors (e.g., size, slant, eyelid architecture, ptosis) can contribute to configuration of the palpebral fissures [Hall et al., 2007].

Note that the plurality of the terms is variable. The default chosen is to specify the singular form of the term unless the term relates to a pair of structures and only makes sense in the plural form (e.g., Eyes, closely spaced) or refers to a structure with many elements (e.g., Eyelashes, sparse). The plurality of the terms was ignored when they were alphabetized and the terms were grouped together (e.g., “Eye...” and “Eyes...” are grouped together, and not interrupted by “Eyelashes...”).

DEFINITIONS

Ablepharon

Definition: Absent eyelids (Fig. 4). objective

Comments: In Ablepharon the globe is continuously exposed [Stevens and Sargent, 2002]. It is arguable whether true aplasia of
the eyelids exists, or whether this represents severe hypoplasia. Nevertheless, as it is difficult or impossible to make this distinction on clinical grounds, the term was felt to be useful.

**Synonym:** Absent eyelids

### Ankyloblepharon

**Definition:** Partial fusion of the upper and lower eyelid margins by single or multiple bands of tissue (Fig. 5).

**Comment:** This term derives from Weiss et al. [1992]. A minimally expressed form, especially when located fully laterally, may be more difficult to ascertain and would be a subjective feature. A band may break and leave no evidence of its presence. Note that this term is distinct from Cryptophthalmos.

**Synonym:** Eyelid synechiae; Ankyloblepharon filiforme adnatum

### Blepharophimosis

**Definition:** A fixed reduction in the vertical distance between the upper and lower eyelids with short palpebral fissures (Fig. 7).

**Comment:** This term is derived from Cunniff et al. [1998]. It is an acknowledged bundled term. When palpebral fissures are severely shortened, they cannot be widely separated, actively or passively. Ptosis is the term to be used when the reduction in eyelid opening is not fixed but can be increased actively or passively. Blepharophimosis is often associated with Epicanthus inversus.

### Cryptophthalmos

**Definition:** Absent palpebral fissures, with skin passing continuously from the forehead or eyebrow onto the cheek (Fig. 8).

**Comments:** This term is based on Saal et al. [1992]. This is an acknowledged bundled term, though the separate coding of the components (palpebral fissure absence; presence of eyelashes) was deemed impractical. This is typically associated with a rudimentary or small globe. Frequently, a tuft of hair accompanies the aberrant skin

### Blepharochalasis

**Definition:** Lax, wrinkled, and baggy eyelid skin (Fig. 6).

**Comment:** This finding is usually more apparent in the upper eyelid. The eyelid tissue thickness is usually also reduced. The feature is frequent in older persons [Held and Schneiderman, 1990].

### Ectropion

**Definition:** An outward turning (eversion) or rotation of the eyelid margin (Fig. 9).
Comment: This term is derived from Cheng and Biglan [2002]. This finding is frequently associated with overexposure of the palpebral and scleral conjunctiva and cornea. It usually involves the lower eyelid. Modifiers such as “lower” and “lateral” may be applied as appropriate.

Entropion

Definition: An inward turning (inversion) of the eyelid margin (Fig. 10). subjective

Comment: The inward turned eyelid margin increases the potential for mechanical irritation of the eye by eyelashes [Cheng and Biglan, 2002]. This should be distinguished from Epiblepharon.

Epiblepharon

Definition: Redundant eyelid skin pressing the eyelashes against the cornea and/or conjunctiva (Fig. 11). subjective

Comment: This term is based on Lemke and Stasior [1981]. This should be distinguished from Entropion (see above).

Epicanthal fold: See Epicanthus

Epicanthus

Definition: A fold of skin starting above the medial aspect of the upper eyelid and arching downward to cover, pass in front of and lateral to the medial canthus (Fig. 12A). subjective

Comment: In extreme cases, the skin fold can start as high as the eyebrow [Hall et al., 2007]. This is called epicanthus superciliaris (Fig. 12B).

Synonym: Epicanthal fold; Epicanthus palpebralis

Epicanthus Inversus

Definition: A fold of skin starting at or just below the medial aspect of the lower lid and arching upward to cover, extend in front of and lateral to the medial canthus (Fig. 12C). subjective

Comment: For additional information on this finding, see [Oley and Baraitser, 1988].

Eyes, Closely Spaced

Definition: Interpupillary distance more than 2 SD below the mean (Fig. 13). objective OR

Interpupillary distance below the 3rd centile. objective OR

The interpupillary distance appears to be decreased. subjective

Comment: The finding is measured according to Hall et al. [2007]. Note that the data from Hall et al. [2007] show SD for newborns and centiles for older patients. In the latter data, there is a continuing increase in this measurement between 14 and 15 years of age, so in the objective finding cannot be made using these norms in persons above 15 years of age. See Figure 3 for a gestalt of normal and abnormal eye spacing.

Synonym: Hypotelorism
Eye, Deeply Set

**Definition:** An eye that is more deeply recessed into the plane of the face than is typical (Fig. 14).

**Comments:** This finding should be distinguished from a prominent supraorbital ridge or inferior orbital margin. In Deeply set eyes, the globe is recessed in comparison to the overall prominence of the face. There is no known objective measurement, and diagnosing this feature depends heavily on the experience of the observer.

**Synonym:** Sunken eyes

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Eyebrow, Broad

**Definition:** Regional increase in width of the eyebrow (Fig. 16).

**Subjective**

**Comment:** broadening or flaring can be medial or lateral, and the term may be modified by appending one of these words to the term (see Fig. 16). Flaring is used to describe a widening with a change in direction of the hairs constituting the eyebrow, but these terms are considered synonymous here.

**Synonym:** Flared eyebrow

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Eye, Widely Spaced

**Definition:** Interpupillary distance more than 2 SD above the mean (newborns 27–41 weeks gestational age Fig. 15). **Objective** OR

Interpupillary distance above the 97th centile (0–15 years of age). **Objective** OR

The interpupillary distance appears to be increased. **Subjective**

**Comment:** The finding is measured according to Hall et al. [2007]. Note that the data from Hall et al. [2007] show continuing increase in this measurement between 14 and 15 years of age, so this finding should only be made according to the subjective definition in persons above 15 years of age. It is important to distinguish between truly increased interpupillary distance and the apparently increased spacing that is caused by Telecanthus (see that entry for additional discussion). See Figure 3 for an illustration of variations of eye spacing.

**Synonym:** Hypertelorism

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Eyebrow, Highly Arched

**Definition:** Increased height of the central portion of the eyebrow, forming a crescent, semicircular, or inverted U shape (Fig. 17).

**Subjective**

**Comment:** Most eyebrows have some arch with down turning medially and laterally. We know of no normative data for eyebrow arching. Identifying this feature is dependent on the experience of the observer. It may help to closely compare the shape/arching with that of siblings and parents.

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Eyebrow, Horizontal

**Definition:** An eyebrow that extends straight across the brow, without curve (Fig. 18).

**Subjective**

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Eyebrow, hirsutism of: See Eyebrow, thick

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Eyebrow, bushy: See Eyebrows, thick

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FIG. 13. Closely spaced eyes, in a child. See also Figure 36.

FIG. 14. Deeply set eyes in a child.

FIG. 15. A boy with Widely spaced eyes, subjective but without Telecanthus, demonstrating the independence of these two features. See also Figures 10, 30, 33, and 40B.

FIG. 16. Broad medial eyebrows in a boy. In this patient, the modifier of medial is inserted into the primary term.

FIG. 17. Highly arched eyebrows in a young man. In this patient, the eyebrows are high-set and thin and they ride above the supraorbital rim, although these features are not required for the term to be used.

FIG. 3. Variations of eye spacing.
**Comment**: Evaluation should be performed with the face at rest. Horizontal eyebrows are an uncommon finding.

**Synonym**: Straight eyebrows

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**Eyebrow, Laterally Extended**

**Definition**: An eyebrow that extends laterally beyond the orbital rim rather than turning gently downward at that location (Fig. 19). subjective

**Comment**: The degree of extension beyond the orbital rim considered abnormal has not been established. This feature is also uncommon.

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**Eyebrow, Sparse**

**Definition**: Decreased density/number and/or decreased diameter of eyebrow hairs (Fig. 20). subjective

**Comment**: Sparseness can be regional (medial, central, lateral) or total. These modifiers should be incorporated into the term, when appropriate.

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**Eyebrow, Thick**

**Definition**: Increased density/number and/or increased diameter of eyebrow hairs (Fig. 21). subjective

**Comment**: Thickness can be regional (medial, middle (central), lateral) or total. These modifiers should be incorporated into the term, when appropriate.

**Synonym**: Hypertrichosis of the eyebrow; Bushy eyebrow

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**Eyelashes, Absent**

**Definition**: No eyelashes are present (Fig. 22). objective

**Comment**: This term is based on Ahmad et al. [1998]. Often this finding is congenital and associated with alopecia universalis, but this should be coded separately.

**Synonym**: Atrichia of eyelashes

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**Eyelashes, Long**

**Definition**: Mid upper eyelash length >10 mm (Fig. 23). objective

**OR**

**FIG. 18.** Horizontal eyebrows, which extend straight across from medial to lateral in this girl. See also Figure 19.

**FIG. 19.** Laterally extended eyebrows in a teenage boy whose eyebrows extend far beyond the lateral orbital wall. He also has Horizontal eyebrows, but this should be coded separately.

**FIG. 20.** Sparse eyebrows in a girl with other features of ectodermal dysplasia. See also Figure 25.

**FIG. 21.** Thick eyebrows, in a patient who also has Prominent eyelashes as well. This pre-teenage boy also shaves his facial hair.

**FIG. 22.** Absent eyelashes or atrichia or in an adult female.

**FIG. 23.** Long eyelashes, in a boy. Note that, in addition to their length, the eyelashes are unusually angled, although that feature is not required for the finding of Long eyelashes. See also Figure 24.
Increased length of the eyelashes. **subjective**

**Comment**: Measurement should be done on the longest lashes, which are usually at the center of the lid. Normal values are 7.99 ± 1.05 mm in boys and 7.76 ± 1.03 mm in girls [Pucci et al., 2005].

**Synonym**: Ciliary trichomegaly.

**Eyelashes, Prominent**

**Definition**: Eyelashes that draw the attention of the viewer due to increased density and/or length and/or curl without meeting the criteria of trichomegaly (Fig. 24). **subjective**

**Comment**: This is admittedly a bundled term, but it may be useful in clinical practice.

**Eyelashes, Sparse**

**Definition**: Decreased density/number of eyelashes (Fig. 25). **subjective**

**Comment**: Sometimes **Sparse eyelashes** are abnormally formed eyelashes. The sparseness may be limited to one portion of the eyelid. We know of no normative data for **Sparse eyelashes**.

**Synonym**: Hypotrichosis of eyelashes

**Eyelid, Cleft**

**Definition**: A short discontinuity of the margin of the lower or upper eyelid (Fig. 26). **subjective**

**Comment**: The lateral segment of the lower eyelid is most commonly involved. As the milder forms of this finding are clearly subjective and no boundary of subjective and objective is defined, the term is considered subjective. The term “eyelid coloboma” has been replaced because the word “coloboma” should be used only for defects at the site of fusion of embryologic structures, which is not the case here. Modifiers to designate the location of the cleft may be added, such as “lower” and “lateral.”

**Synonym**: Notched eyelid

**Replaces**: Eyelid coloboma

**Infra-Orbital Crease**

**Definition**: Skin crease extending from below the inner canthus laterally along the malar process of the maxilla and zygoma (Fig. 27). **subjective**

**Comment**: This feature is often found in the presence of hypoplasia of the malar process of the maxilla or zygoma, but this should be described separately. See **Infra-orbital fold** for a related term.

**Infra-Orbital Fold**

**Definition**: Elevated ridge(s) of skin starting well below the medial aspect of the lower lid that curves gradually upward toward and/or across the nasal bridge (Fig. 28). **subjective**

**Comments**: This excludes **Epicanthus inversus**. See **Infra-orbital crease** for a related term.

**Lacrimal Punctum, Absence**

**Definition**: No identifiable superior and/or inferior lacrimal punctum (Fig. 29). **objective**

**Comments**: The openings of the tear ducts are normally located at the medial margin of each eyelid. The opening on the lower eyelid
border is more visible than the opening on the upper eyelid border. Absence of the lacrimal punctum is uncommon [Ferreira et al., 2000] and although it is typically presumed to be caused by agenesis, we avoid causal mechanisms in this terminology. There was some disagreement as to whether Nasolacrimal duct obstruction should be included in this terminology. As that assessment is primarily functional and not anatomic, it was removed.

Replaces: Agenesis of the lacrimal punctum

Lacrimal Punctum, Ectopic

Definition: Positioning of a lacrimal punctum other than at the medial margins of the eyelid (Fig. 30). subjective

Comment: The openings of the tear ducts are normally located at the medial margin of each eyelid. The opening on the lower eyelid border is more visible than the opening on the upper eyelid border. Ectopic positions can include the upper eyelid, nasal bridge, or inferior to medial aspect of the lower lid.

Palpebral Fissure, Almond-Shaped

Definition: A shape created by an acute downward arching of the upper eyelid and upward arching of the lower eyelid, toward the medial canthus, which gives the outline of the palpebral fissures the configuration of an almond; thus, the maximum distance between the fissures is offset from, and medial to, the center point (Fig. 32). subjective

Comments: The almond configuration tends to dissipate with time as the surrounding tissues (e.g., eyelid, nasal bridge) grow.

Palpebral Fissure, Downslanted

Definition: The palpebral fissure inclination is more than 2 SD below the mean for age (Fig. 33). objective OR subjective

Comments: The slant, or inclination, of the palpebral fissure is less than typical for age.

Lagophthalmos

Definition: Inability to totally close the eyelids while awake, asleep, or both (Fig. 31). subjective

Comment: It may be an isolated finding or part of a syndrome [Korula et al., 1995] and can be associated with Ectropion. Lagophthalmos frequently results in chronic conjunctival and/or corneal irritation.

Mongoloid slant: See Palpebral fissure, upslanted

Palpebral Fissure, Upslanted

Definition: The palpebral fissure inclination is more than 2 SD above the mean for age (Fig. 34). subjective OR subjective

Comments: The slant, or inclination, of the palpebral fissure is greater than typical for age.

Infra-orbital folds associated with upper facial edema [the latter is not required for the finding to be made]. Note that these folds are oriented toward the lateral borders of the nasal bridge. See also Figure 27.

This child has Absence of the lacrimal puncta. Note that she also has Short palpebral fissures, Downslanted palpebral fissures, Telecanthus, and Nasolacrimal duct obstruction with erythematous and swollen lacrimal ducts below the medial canthi.

The left eye shows an inferiorly placed medial canthus and Ectopic lacrimal punctum plus a defect in the left nostril.
speciﬁed in Farkas [1994] for Caucasians (pg. 283) and for Chinese and African-American populations (limited data, pgs 342, and 349, respectively). Hall et al. [2007] only speciﬁes norms for Caucasians between 6 and 16 years of age. Note that the mean inclination is slightly upslanting at all ages [Farkas, 1994; Hall et al., 2007]. Some features (e.g., Ptosis or Epicanthus) may hinder palpebral ﬁssure inclination assessment. Malar and/or zygomatic hypoplasia and Widely spaced eyes may be associated with a downward slant.

Replaces: Antimongoloid slant

Palpebral Fissure, Long

Definition: Distance between the medial and lateral canthi is more than 2 SD above the mean for age (Fig. 34). objective OR

Apparent increased length of the palpebral fissures. subjective

Comment: Measurement techniques and norms are as speciﬁed in Hall et al. [2007] and Farkas [1994]. The term “wide palpebral fissure” is discouraged because this term is vague with respect to which dimension (vertical vs. horizontal) it refers to, coupled with the popular deﬁnition of “wide-eyed,” which refers to palpebral fissure height.

Replaces: Wide palpebral fissure

Palpebral Fissure, Short

Definition: Distance between the medial and lateral canthi is more than 2 SD below the mean for age (Fig. 35). objective OR

Apparently reduced length of the palpebral fissures. subjective

Comment: Measurement techniques and norms are as speciﬁed in Hall et al. [2007] and Farkas [1994]. These sources include more complete data for Caucasians than for other groups. Decreased palpebral fissure length may be accompanied by a decreased vertical distance between the upper and lower eyelid, which gives the eyes a more slit-like appearance and may give the appearance of Ptosis, but this should be coded separately if it is present. Severe shortening of the palpebral ﬁssures leads to Blepharophimosis.

Palpebral Fissure, Upslanted

Definition: The palpebral ﬁssure inclination is more than 2 SD above the mean for age (Fig. 36). objective OR

The inclination of the palpebral ﬁssure is greater than typical for age. subjective

Comment: The slant, or inclination, of the palpebral ﬁssure is deﬁned as the angle formed by two lines: an imaginary line that connects the lateral canthus and the medial canthus of each eye, and an imaginary horizontal line formed by the two medial canthi when the patient holds their head with the facial midline vertical, the head in a neutral vertical position (neither ﬂexed nor extended) and the gaze forward [Farkas, 1994]. Palpebral ﬁssure inclination norms are speciﬁed in Farkas [1994] for Caucasians (pg. 283) and for Chinese and African-American populations (limited data, pgs 342, and 349, respectively). Hall et al. [2007] only speciﬁes norms for Caucasians between 6 and 16 years of age. Uplanted palpebral ﬁssures may be associated with microcephaly, but this should be coded separately. Some features (e.g., Ptosis or Epicanthus) may hinder palpebral ﬁssure inclination assessment.

Replaces: Mongoloid slant

Proptosis

Definition: An eye that is protruding anterior to the plane of the face to a greater extent than is typical (Fig. 37). subjective
Comments: This finding should be distinguished from underdevelopment of the supraorbital ridge or maxilla/zygoma. In Proptosis, the globe is anteriorly protuberant in comparison to the overall plane of the face. There is no known objective measurement, and diagnosing this feature depends heavily on the experience of the observer.

Synonym: Prominent eyes

Ptosis

Definition: The upper eyelid lid margin is positioned 3 mm or more lower than usual and covers the superior portion of the iris (Fig. 38). Objective OR

The upper lid margin obscures at least part of the pupil. Subjective Comment: True ptosis usually occurs in the presence of normal palpebral fissure length. Apparent ptosis is seen in Blepharophimosis and other causes of Short palpebral fissures. Pseudoptosis occurs in the presence of severe zygomatic underdevelopment when the resulting dramatic downward eye slant pulls the upper eyelid diagonally across the globe.

Synonym: Blepharoptosis

Synophrys

Definition: Meeting of the medial eyebrows in the midline (Fig. 39). Subjective

Comment: Cosmetic hair removal or shaving may obscure this feature. It is controversial whether the medial eyebrows must meet in the midline to warrant this descriptor, as opposed to eyebrows that extend markedly toward the midline but do not meet.

Replaces: Unibrow

Telecanthus

Definition: Distance between the inner canthi more than 2 SD above the mean (Fig. 40). Objective OR

 Apparently increased distance between the inner canthi. Subjective

Comments: Telecanthus may be present without (Fig. 40A) or with (Fig. 40B) Widely spaced eyes. In the latter case, Widely spaced eyes should be coded separately. Inner canthal distance varies among ethnic groups. Norms are available for American Africans [Murphy and Laskin, 1990], Chinese [Wu et al., 2000], and Caucasians [Laestadius et al., 1969; Feingold and Bossert, 1974; Merlob et al., 1984;Evereklioglu et al., 2001]. In the presence of an Epicanthal fold, ascertainment of inner canthal distance can be difficult.

Synonym: Dystopia canthorum

Synchiae: See Ankyloblepharon

Unibrow: See Synophrys

Upper Eyelid, Fullness of

Definition: Swelling or distention of the upper eyelid (Fig. 41). Subjective
Comment: The swelling can be due to edema, fat or other deposits or inflammation. It is controversial whether there should be a separate term for fullness of the lateral upper eyelid.

REFERENCES


