INIVITED COMMENT

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# A User's Guide to the Elements of Standard Morphologic Terminology: Analysis and Database

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The morphologic examination combines subjective assessment with objective measurement, approaches that are systematized by Elements of Morphology: Standard Terminology [EMST09—Carey, 2009; Allanson et al., 2009a]. This work derived from previous efforts [Spranger et al., 1982—IWG82] and was produced over 4 years by a consortium of 34 clinicians in 6 committees (designated here also as an International Working Group—IWG09). A need to assimilate EMST09 terms and definitions, and facilitate review prompted me to create a Microsoft Excel database (available upon request). My commentary serves as informal dysmorphologist's "user's guide" on the coverage and precision of EMST09, with suggestions to promote its use in clinical practice and research.

The remarkable scope of IWG09 is summarized in Table I including 394 morphologic terms defined by the six region-oriented subcommittees. I entered all terms, but condensed some definitions and comments, grouping these elements into 39 subregions. Of these terms, 236 (60%) are scored by IWG09 as subjective and 158 (40%) as objective; those scored as objective include 91 recognized by observation, 44 specified by >2 SD from age-matched norms using a tape measure, 6 specified by formulas (e.g., cephalic index) using tape measurements, and 17 requiring measure using calipers, angles, or special instrument (e.g., palatal height).

There are 71 terms (18% of total) defined for the head and face [Allanson et al., 2009b] with 10 for the cranium, 9 for the face overall (e.g., round face), and 12–14 for the upper, mid (maxillary), or lower (mandibular) facial regions (Table I). Similar descriptive detail is accomplished for the periorbital region [39 terms: 10%; Hall et al., 2009], nose/philtrum [51 terms: 12.9%; Hennekam et al., 2009], ear [74 terms: 18.7%; Hunter et al., 2009b], oral region [58–14.7%; Carey et al., 2009], and hands/feet [101–25.6%; Biesecker et al., 2009]. Although extensive, the coverage is incomplete as discussed by Allanson et al. [2009b] in their introduction and reiterated by Hunter and Hennekam [2009] in response to the comment by Klinger and Merlob [2009]. These authors suggested adding several terms for ear morphology such as "antihelix, superior crus, absent" which has been added to the database, an inadvertent omission according to Hunter and Hennekam [2009]. This illustrates the dynamic nature of the EMST09, and supports the IWG09 vision for a morphologic nomenclature committee [Allanson et al., 2009a]. EMST should become a dynamic resource able to correct a few inconsistencies or omissions, and issue periodic updates. Extension to other surface anomalies of the skin, back, and genitalia (e.g., "ash-leaf spot," "narrow areolae," and "sacral dimple") and re-evaluation of IWG82 terminology (e.g., "syndrome," "disruption") would be extremely valuable.

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Analysis of individual definitions supported the precision of IWG09 work, which rejected "bundled" terms like "hypotonic," "aged," or "immobile" face that compound several morphologic elements. The restriction to altered form was a necessary first stage, exemplified by omission of terms such as "hypertrophy" or "hypogenesis" [Hunter and Hennekam, 2009; Klinger and Merlob, 2009]. Since morphologic variations provide evidence of dysfunction that is essential to causal analysis (e.g., "narrow forehead" and "down-turned corners of mouth" with hypotonia), inclusion of these terms is an important justification for continued IWG activity. The precision of IWG09 definition is also exemplified by listing allowable synonyms (e.g., "furrowed tongue" as preferred term with "prominent tongue grooves" as synonym) and by excluding certain terms [e.g., "scrotal tongue" is replaced because it is pejorative—Biesecker et al., 2009]. My database italicizes synonyms and terms to be replaced and places them in a separate column.

Other users may share my impression that EMST09, by virtue of its precision and breadth of coverage, provides a reference for morphologic variation akin to those for chromosomal or DNA sequence variations [Carey, 2009]. In the future, attention can be given to:

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TABLE I. Summary of Morphologic Elements by Region* and Subregion**

| Section | Symbol | Total | Subj | Obj-O | Obj-T | Obj-TF | Obj-SI |
|---|---|---|---|---|---|---|---|
| Head and face [Allanson et al., 2009b] | HF | 71 (18%) | 52 | 3 | 7 | 2TF | 8 |
| Cranial | HF-Cr | 10 | 5 | 0 | 3 | 2TF | |
| Hair | HF-Ha | 9 | 5 | 2 | 2 | 0 | |
| Face | HF-Fa | 9 | 5 | 0 | 2 | 0 | 3C |
| Forehead | Hf-For | 12 | 9 | 0 | 0 | 0 | 2C 1A |
| Maxillary region | HF-Max | 12 | 12 | 0 | 0 | 0 | |
| Mandibular region | HF-Jw | 14 | 11 | 1 | 0 | 0 | 2C |
| Neck | HF-N | 5 | 5 | | | | |
| Periorbital region [Hall et al., 2009] | | 39 (10%) | 26 | 5 | 6 | 0 | 4 |
| Eye periorbital region | EY-PER | 6 | 3 | 0 | 3 | 0 | 0 |
| Eye brows | EY-Bro | 7 | 7 | 0 | 0 | 0 | 0 |
| Eye palpebral fissues | EY-PF | 8 | 2 | 2 | 2 | | 2A |
| Eye lids | EY-LID | 14 | 12 | 2 | 0 | 0 | 0 |
| Eye lashes | EY-Las | 4 | 2 | 1 | 1 | 0 | 0 |
| Nose and philtrum [Hennekam et al., 2009] | NP | 51 (12.9%) | 33 | 9 | 9 | 0 | 0 |
| Nose, entire | NP-No | 11 | 3 | 3 | 5 | 0 | 0 |
| Nose-bridge, ridge | NP-NoSup | 9 | 8 | 1 | 0 | 0 | 0 |
| Nose tip | NP-NoTip | 6 | 6 | 0 | 0 | 0 | 0 |
| Nose nares | NP-NoNar | 5 | 4 | 1 | 0 | 0 | 0 |
| Nose base | NP-NoBas | 10 | 8 | 2 | 0 | 0 | 0 |
| Nose philtrum | N-Ph | 10 | 4 | 2 | 4 | 0 | 0 |
| Ears [Hunter et al., 2009b] | | 74 (19.2%) | 34 | 33 | 5 | 0 | 2 |
| Ear pinna | Er-Pin | 14 | 3 | 4 | 5 | 0 | 2A |
| Ear helix | Er-Hel | 19 | 11 | 8 | 0 | 0 | 0 |
| Ear pattern (antihelix, tragus, concha) | Er-Pat | 33 | 17 | 16 | 0 | 0 | 0 |
| Ear lobe | Er-Lob | 8 | 3 | 5 | 0 | 0 | 0 |
| Oral region [Carey et al., 2009] | LM | 58 (14.7%) | 31 | 14 | 8 | 2 | 3 |
| Perioral region | LM-PerOr | 3 | 3 | 0 | 0 | 0 | 0 |
| Mouth | LM-Mo | 4 | 2 | 0 | 2 | 0 | 0 |
| Lips | LM-Lip | 13 | 6 | 3 | 4 | 0 | 0 |
| Gums | LM-Gum | 6 | 4 | 2 | 0 | 0 | 0 |
| Teeth | LM-Tth | 12 | 3 | 5 | 2 | 2F | 0 |
| Tongue | LM-Tng | 9 | 8 | 1 | 0 | 0 | 0 |
| Palate-uvula | LM-Pal | 11 | 5 | 3 | 0 | 0 | 3I |
| Hands and feet [Biesecker et al., 2009] | HF | 101 (25.6%) | 63 | 27 | 9 | 2 | 0 |
| Hands | HF-H | 11 | 6 | 1 | 4 | 0 | 0 |
| Feet | HF-F | 15 | 6 | 5 (2M) | 4 | 0 | 0 |
| Digits | HF-D | 4 | 2 | 2 | 0 | 0 | 0 |
| Hand digits (fingers) | HF-HD | 28 | 19 | 7 | 1 | 1M | 0 |
| Hand digits (thumbs) | HF-HTh | 7 | 2 | 4 (1M) | 0 | 1TF | 0 |
| Foot digits (toes) | HF-FD | 14 | 10 | 4 | 0 | 0 | 0 |
| Foot digits (halluces) | HF-FHal | 3 | 2 | 1 | 0 | 0 | 0 |
| Hand creases | HF-HC | 6 | 3 | 3 | 0 | 0 | 0 |
| Foot creases | HF-FC | 1 | 1 | 0 | 0 | 0 | 0 |
| Nails | HF-N | 12 | 12 | 0 | 0 | 0 | 0 |
| Totals | | 394 | 239 | 91 | 44 | 6 | 17 |

*Defined by EMST09 [Carey, 2009] or **arbitrarily by the author; anomaly detected by Subj, subjective; Obj-O, objective observation with manipulation (M); Obj-T, objective by tape measure as >2 standard deviations from mean; Obj-TF, objective by tape measure and/or special formula (F); Obj-SI, objective by measure with calipers (C), angle (A), or other instrument (I).

(1) Improve consistency for some terms that include variable use of "breadth, width, girth" with "broad" more usual than "wide"—for example, "wide nasal base" and "wide nasal bridge" when "broad" is employed in other terms (e.g., "broad face, forehead, hallux, thumb, toe"). Use of a synonym is appropriate in definitions but consistency is needed for the terms themselves. Terms such as "alveolar ridge overgrowth" or "gingival overgrowth" imply mechanism—would not "broad alveolar ridge" be more consistent with the intent to avoid functional description?
(2) Improve clarity for some definitions or comments, for example, comment for "thumb, proximal placement of."

Comments for several terms ask the examiner to specify digits (e.g., 2, 4–5); this instruction could be more economically stated in the section introduction. Many Latin terms like "micronychia," "brachynychia," or "pes planus" are retained where simple English might be considered.

(3) Triage the 394 terms in EMST09 to encourage widespread clinical usage, perhaps at two levels: Devise condensed but relatively detailed morphologic assessment scales for genetic and developmental subspecialists while selecting those elements deemed most definitive and frequent for generalists. Record individual elements by region and sum their total, exercising care not to equate any numerical score with a specific conclusion (e.g., "dysmorphic" or not without considering family background). These morphologic assessment scales could be associated with CPT codes analogous to those for assessment of child development. Examinations adapted and endorsed by IWG could be billed as additions/modifications to the 99241-5 or 99251-5 for outpatient/inpatient genetic/developmental consultation, thus promoting appreciation of morphologic expertise and recompense for time. Analysis of structural variations in particular disorders might assist triage, as performed for the ear in Brachmann-de Lange syndrome [Hunter et al., 2009a]. Extension of their study to disorders like trisomy 18 or branchio-oto-renal syndrome might establish correlations among ear size/ear pattern elements that would condense the 74 ear descriptors (Table I).

(4) Refine morphologic terminology in concert with IWG meetings and revisions, updating prior epidemiologic surveys [Mehes, 1985; Leppig et al., 1988] and initiating new ones using EMST09 terms and definitions. Such studies could assist triage by updating anomaly frequencies and assess the quality and time requirements of morphologic examinations. Serial assessments by examiners with varied experience could define sensitivity and reproducibility of results, including incidences of instrument injuries, stress to children, and reactions of parents.

Beyond its obvious benefits for clinical delineation and scholarship, EMST09 provides a sort of periodic table for future research. Facial recognition by patients with prosopagnosia, autism, or mental illness [Bowles et al., 2009] could be studied as a function of the variable elements defined by EMST09. The role of component regions in facial recognition may provide insights into maternal–infant bonding, discrimination of friend or foe, and thus, into human social evolution [DeBruine et al., 2008]. Early applications could improve the focus of computerized identification systems [Castillo and Jacobs, 2009], while long-term studies could examine genotype–phenotype correlations among calibrated facial elements during primate evolution [O'Higgins, 2000; Ou et al., 2008].

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