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REVIEW ARTICLE

FACIES IN DERMATOLOGY

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ABSTRACT

Economic Faces are but a gallery of pictures. "-Francis Bacon. Facies is the appearance of a facial expression of an individual. In the deviations from the normal facies lie subtle clues. These clues may point the keen observer not just to myriad possibilities from the coarse facies of inborn errors of metabolism to leonine facies, but at times can be the tell-tale symptom of the disease itself like syphilitic facies. Nevertheless the distinct appearance holds the clue to the diagnosis and is an integral part of clinical examination. This article brings in together the descriptions of various facies to not only emphasize but also categorise the same for the sake of convenience of the observant.

INTRODUCTION

"Evil was coming. I wondered whose face it would be wearing." This liner from a famous book has medical implications too! It is fascinating how different morbidities manifest with typical pathognomic facial features that make the diagnosis so obvious. Some are pre-emptive to the disease fulminance (cadaveric facies), some indicative of the incumbent criticality (facies of cirrhosis of liver) while the remaining represent the signatures of complications endured (congenital syphilis). Some are resultant to infections (leprosy), some to endocrinal (thyrotoxic or myxeodematous facies) and metabolic disturbances (muccopolysaachidoses, galactosialidosis) while yet some are subsequent to drug use (phenytoin). Many fascinating names like bovine facies (Crouzon syndrome), hatchet facies (Myotonia atrophica), elfin facies (William's syndrome), leonine facies (lepromatous leprosy), bird facies (Pierre Robin malformation) and chipmunk facies (beta thalassemia major), etc have been used to describe these. Names suggestive of etiology have also been used (amiodarone facies, adenoid facies, etc). An understanding of the obvious and the intricate features of the facies makes for an interesting journey through the lanes of clinical medicine and human imaginations. This article enumerates some of the common, fascinating and typical facies one may come across in day to day practice as a dermatologist/ physician or a general practitioner.

Some of the important diagnostic facies are

- Acromegalic facies (Acromegaly): Large supraorbital ridge and frontal bossing, protruding, oedematous thick eyelids, triangular large ears, numerous skin tags ('fibroma molluscum'), widely spaced teeth, enlarged and furrowed tongue, , thickened lower lip, lower jaw firm and square (protruding jaw = prognathism) and cutis gyrata of the scalp in more extreme cases. Periosteal new bone formation of the facial bones and skin causes the characteristic facies[1.]
- Asciatic porcelain doll facies: Restrictive dermopathy (RD) is characterized by abnormal facies, tight skin and secondary joint changes that are diagnostic. The skin is shiny and taut. The face is typical with a small, fixed and round open mouth, micrognathia, small nose and widely set ears. The face is expressionless with blurring of groove between nose and cheek and sparse or absent eyelashes, best described as 'Asiatic porcelain doll" [2].
- Facies of acnathosis nigricans: MORFAN syndrome[3] (mental retardation, overgrowth, remarkable facies with extreme pigmentation, and acanthosis nigricans)
- Adenoid facies (Adenoid hypertrophy): Long, open-mouthed, dull and dumb-looking face of children with hypertrophy of pharyngeal tonsils (adenoids) [4]. Cowden's syndrome syn. Multiple hamartoma syndrome characterised by skin-coloured lichenoid papules, that tend to coalesce to give a cobblestone appearance, distributed on and around the eyes and mouth, is associated with adenoid facies. Carney complex is associated with adenoid facies[5].
- Facies of addison's disease: Generalized darkening of the skin of face along with pigmentation in the mucous membranes within the mouth.
- Amiodarone facies: Deep blue discoloration around malar area and nose [6].
- Antonine facies: in tuberculoid leprosy, involvement of facial nerves produces a blank expression known as antonine facies [7].
- Facies of argyria: Seen amongst workers in silver. The coloration is even and uniform with blue-grey appearance and persists when pressure is applied to the skin. It is subcutaneous rather than dermal pigmentation. The condition is also known as pseudo ochronosis [8].

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- Bell's palsy (Facial nerve dysfunction): The eyelids on the paralyzed side can't close. The mouth is drawn to the unparalyzed side, producing a somewhat grotesque appearance. Food and drink dribble from the mouth on the paralyzed side. The eye with the involved lid dries due to decreased tear production.
- Bird facies (Pierre Robin Malformation): Small lower jaw, a slit like hole in the palate of mouth (called cleft palate) and the tongue appear to fall into the throat (condition called as retroglossoptosis)[9]. Also seen in Mulvihill–smith syndrome, Treacher Collins syndrome, Goldenhar syndrome, Nager syndrome, Miller syndrome[10].
- Bovine facies (Craniofacial Dysostosis or Crouzon syndrome): Convex nasal profile, shortened mandible, macroglossia
- Coarse facies[11] describes a constellation of facial features that are present in many inborn errors of metabolism. Features include: large, bulging head prominent scalp veins "saddle-like, flat bridged nose with broad, fleshy tip" large lips and tongue small, widely spaces and/or malformed teeth, hypertrophic alveolar ridges and/or gums. Heads tend to be longer than normal from front to back, with a bulging forehead. This is because of the earlier than normal or premature fusion of skull bones in an affected individual. Causes :

1. Alpha-mannosidosis type II
2. Aspartylglycosaminuria
3. Battaglia Neri syndrome
4. Borjeson Syndrome
5. Chromosome 6q deletion syndrome
6. Coarse face - hypotonia - constipation
7. Congenital hypothyroidism
8. Dandy-Walker malformation
9. Dyggve-Melchior-Clausen Syndrome
10. Fucosidosis type 1/2
11. Gangliosidosis generalized GM1 (type 1)
12. Gangliosidosis GM1 (type 3)
13. GM1 gangliosidosis
14. Goldberg syndrome
15. Hyde-Forster-Mccarthy-Berry syndrome
16. Hyper IgE
17. Hypomelanosis of Ito
18. Immunodeficiency due to defect in MAPBP-interacting protein
19. Infantile sialic acid storage disorder
20. Job syndrome
21. Mannosidosis (alpha B lysosomal)
22. McCune-Albright Syndrome
23. Mental retardation (X-linked - epilepsy - progressive joint contractures - typical face)
24. Mental retardation (X-linked Raynaud type)
25. Miescher's syndrome
26. Morquio syndrome
27. Morquio syndrome type A/B
28. MPS 3 C/D
29. Mucolipidosis III
30. Mucopolysaccharidosis
31. Multiple endocrine abnormalities
32. Neuraminidase deficiency (type II juvenile form)
33. Nodulosis-arthropathy-osteolysis syndrome
34. Nonkeratan-sulfate-excreting Morquio syndrome
35. Pituitary tumors (adult)
36. Sialidosis type II (congenital/infantile)
37. Sialuria syndrome
38. Simpson-Golabi-Behmel syndrome
39. Skeletal dysplasia - coarse facies - mental retardation
40. Spondyloepimetaphyseal dysplasia (genevieve type)
41. Sulfatidosis juvenile (Austin type)
42. Winchester syndrome

- Assymeteric crying facies (Cayler cardiofacial syndrome): Asymmetric appearance of the oral aperture and lips at rest, but significant depression of one side of the lower lip with animation (crying or smiling) [12]
- Cadaveric facies : Slow symmetrical disappearance of the facial fat in patients of lipodsytrophy , producing a cadaverous appearance
- Chipmunk facies (B-Thalassemia major, Bullimia nervosa, Parotid swelling): Expanded globular maxillae, with BM hyperexpansion into facial bones, combined with prominent epicanthal folds[13, 14].
- Cushingoid facies (Cushing syndrome): A moon face and hirsuitism with a double chin, prominent flushed cheeks, and fat deposits in the temporal fossa and cheek are characteristic often seen as a result of corticosteroid therapy.
- Facies of dermatomyositis: A purplish red heliotrope erythema occurs on face especially involving the eyelids, upper cheeks, forehead and temples. Odema of eyelids and periorbital tissues is also seen [15].
- Dog or simian facies: Traditionally, cases of congenital hypertrichosis have been classifi ed into two groups—'dog faced' and 'simian[16]. The hair gradually lengthens until by early childhood the entire skin, apart from the palms and soles, is covered by silky hair, which may be 10 cm or more long. Long eyelashes and thick eyebrows are conspicuous features.
- Elfin facies (William's Syndrome): Sunken nasal bridge, puffiness around eyes, epicanthal fold, blue starry eyes, long upper lip length, small and widely spaced teeth, small chin [17].
- Gaunt facies: A common cutaneous side effect of HAART is facial lipoatrophy. The hollowed out cheeks, temples and eye sockets often lead to a gaunt cachetic facies[18] which can be a disconcerting stigmata of the disease and a psychological burden to the patient.
- Gargoyle facies (Hurler syndrome and Galactosialidosis): Head is large and dolichocephalic, with frontal bossing and prominent sagittal and metopic sutures, with mid-face hypoplasia, depressed nasal bridge, flared nares, and a prominent lower 1⁄3 of face, thickened facies, widely spaced teeth and attenuated dental enamel, gingival hyperplasia[19].
- Hippocratic facies: A pinched expression of the face with sunken eyes concavity of cheeks and temples, relaxed lips and leaden complexion seen in one close to death after severe and prolonged illness
- Hound dog facies: The facial appearance in cutis laxa called "bloodhound facies" an aged appearance with sagging jowls with downward slanting palpebral fissures, a broad flat nose and large ears[20].
- Facies of Hartnup disease: *Hartnup disease* is a rare inborn error of metabolism with autosomal recessive inheritance characterized by pellagra like cutaneous eruptions, neurologic abnormalities and a specific amino-aciduria. Cerebellar ataxia is a prominent feature which manifests as unsteady, wide-based gait[21].
- Keel like facies: seen in Bloom's syndrome patients bear a striking resemblance to each other. They have a narrow, slender, delicate facies with a relatively prominent nose and high-arched palate. The essential features are erythema of the face and stunted growth (both prenatal and postnatal) [22].
- Leonine facies : Peculiar, deeply furrowed forehead and cheek, lionlike appearance of the face. Seen in lepromatous leprosy, chronic actinic dermatitis, parthenium dermatitis, papular mucinosis, multricentric reticulohistiocytosis, pseudoly mphoma, mycosis fungoides, scleromyxedema, chronic lympho dema as a complication of rosacea, Carcinoid syndrome, Focal facial dermal dysplasia, KID syndrome (keratitis, ichthyosis, deafness), Leishmaniasis, Lipoid proteinosis, Lymphoma, leukaemia, Mycosis fungoides, Multiple keratoacanthoma syndrome, Pachydermoperiostosis/cutis verticis gyrate, Paget's disease of bone, Progressive nodular histiocytosis, Pseudophotodermatitis

- Facies leprosa: atrophy of anterior nasal spine and maxillary alveolar process contributes to nasal collapse and loss of upper central incisor teeth or of all four upper incisors respectively and these two skull changes have been given the name 'facies leprosa' [23].
- Facies Lactrodectismica (Latrodectus facies): seen with black widow spider envenomation. Facial swelling, painful frimace, flushing, diaphoresis, trismus and blepharitis [24].
- Facies of lupus erythematosus: A confluent symmetrical bluish eruption with fine scaling and odema centred over malar eminence with inflammation extending over bridge of nose completes the body of classic butterfly seen in acute cutaneous lupus erythematosus[25].
- Facies of cirrhosis of liver :

Figure 1. Facies in systemic sclerosis

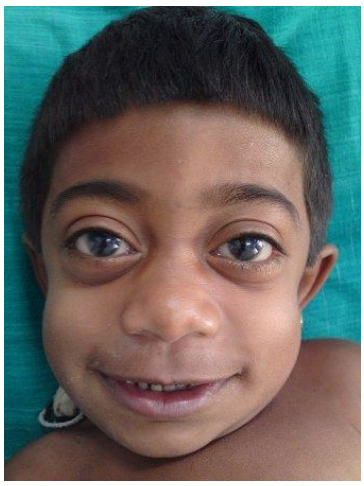

Figure 2. Coarse Facies in Mucopolysaccharidosis

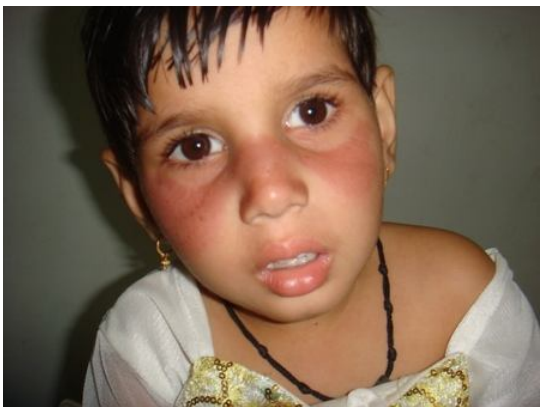

Figure 3. Pellagra type rash in Hartnup's disease

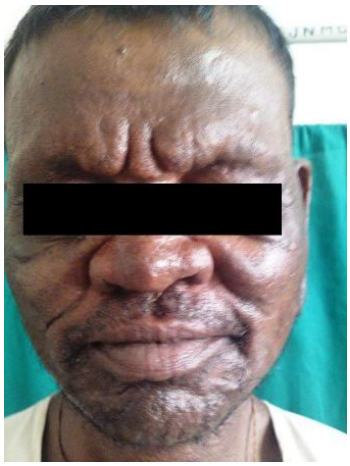

Figure 4. Leonine Facies in Lepromatous Leprosy

i. Early stage: telangiectases over the cheek, coarsening of tissues, especially on and around nose and mouth with purplish reddening in general.
ii. Later stage: sallow, dull diffusely pigmented facies is often distinctive.

- Mongolian facies: Head is brachycephalic, palpebral fissures slant obliquely inwards and downwards towards a broad flat nose, rendered even broader by the presence of epicanthus. Ears are larger and pitcher shaped; the lips are fissured and often left open to allow coarse tongue to protrude; forehead is downy, and the hair of the scalp scanty, wiry, and frequently mouse-colored; the complexion is florid and mottled.
- Monkey like facies: Seen in porphyria cutanea tarda [8].There is hypertrichosis of face, involving cheeks, temples, and eyebrows giving a monkey-like facies.
- Facies in MEN 2B Syndrome: Usually there are numerous yellowish-white, sessile, painless nodules on the lips or tongue, with deeper lesions having normal coloration. There may be enough neuromas in the body of the lips to produce enlargement and a "blubbery lip" appearance. Similar nodules may be seen on the sclera and eyelids.
- Myopathic facies seen in cerebrotendinous xanthomatosis syn. cholestanolosis : An expressionless face with sunken cheeks and a drooping lower lip characteristic of patients with myopathies, especially myotonic dystrophy, associated with open mouth and protruberant tongue
- Micky mouse facies: seen in COCAYNE'S SYNDROME[26] characterized by Facial erythema in a butterfly distribution with mottled pigmentation and atrophic scars. There is loss of subcutaneous fat on face and the sunken eyes gives premature senile appearance. The association of these features with large protruding ears gives a fanciful resemblance to micky mouse.
- Naevoid basal cell carcinoma syndrome syn. Basal cell naevus syndrome; gorlin's syndrome: A highly characteristic facies [27] (broad nasal root, hypertelorism, frontal bossing), jaw cysts , bifid or otherwise misshapen ribs, vertebral and other skeletal anomalies, pits of the skin of the palms and soles, dysgenesis of the corpus callosum, calcification of the falx cerebri (at an earlier age than is seen in non-affected individuals) and macrocephaly. Characteristic facies may occur due to increased calvarial size.
- Facies in noonan syndrome [37]: Patients are of short stature with broad short webbed neck, the facies shows a characteristic association of hypertelorism, blepharoptosis, epicanthic folds and a small chin, widespread leukokeratosis of the lips and gingiva, coarse, light coloured and curly hair, with a low posterior hairline. Downy hypertrichosis may occur on the cheeks or shoulders.

- Flat facies (Down syndrome) [37]: Flat appearing face, small head, flat bridge of the nose, smaller than normal, low-set nose, small mouth which causes the tongue to stick out and to appear overly large, upward slanting eyes, epicanthal fold, rounded cheeks, small misshapen ears.
- Parkinsonian facies (Parkinsonism): Mask-like, tremor of head, absence of blinking, dribbling of saliva, weakness of upward gaze, seborrhoea and sweatiness.
- Phenytoin facies: Patients who use in the long-term commonly manifest with gingival hyperplasia, coarsening of the facies, and hirsutism.
- Facies in premature ageing syndrome[28] :

a.pangeria
b.progeria
c. acrogeria

a.Pangeria: Beaked nose, skin of ears is atrophic and tightly bound down. Skin is dry atrophic, mottled hyperpoigmentation and telangiectasias

b.Progeria : Facial appearance is reminiscent of a fledging bird, with a disproportionately large cranium with patent frontanelles, frontal bossing, prominent eyes and scalp veins, very sparse scalp hair, sparse or absent eyebrows and eyelashes, centrofacial cyanosis, micrognathia, thin lips and a "beaked nose" [29].

c.Acrogeria: Face appears pinched with a hollow cheeked owl eyed appearance a beaked nose and thin lips. Micrognathism may be present. The loss of subcutaneous fat accentuates the appearance of premature senility.

- Raccon facies: erythematous scaly eruption on face and periorbital skin in patients with neonatal systemic lupus erythematosus. Also seen in primary systemic amyloidosis [30] following prolonged dependent positioning in a proctologist's chair, the skin surrounding the eyes shows striking petechiae formation, resembling the mask-like facies of a raccoon.
- Rounded or asymmetrical facies: Seen in X-linked dominant ichthyosis Syn. Conradi–hünermann–happle syndrome along with frontal bossing, a broad flat nasal bridge, and congenital asymmetric cataracts in 60% of patients.
- Snarling or Myasthenic facies (Myasthenia gravis): Drooping of the eyelids and corners of the mouth and weakness of the facial muscles.
- Sad man facies: seen in staphylococcal scalded skin syndrome[31]. Patients demonstrate sad man facies, perioral crusting and radial fissuring with mild facial edema.
- Slapped cheek facies: in erythema infectiosum diffuse erythema gives slapped cheek facies appearance[32].
- syphilitic facies is another separate group: please make it as

a.stokes facies
b.tabetic fcies
c.bull dog facies
d.old man facies[33:]

a.Stokes facies: Stokes has described the facial appearance of many congenital syphilis who may lack the gross stigmata in the following words: 'The essence of matter lies in a certain inalertness, a sleepy, tired, fagged, clouded, dreaming or obscured appearance of the upper face, an appearance of veildness as if a smudge had lightly swept across the brows and eyes and the nasal bridge of a crayon portrait.'

b.Tabetic facies: Argyll Robertson pupils, dropping of upper eyelids due to hypotonia with some wrinkling of forehead imparting a sad expression.

c.Bulldog facies: Frontal bossing, saddle nose and bulldog jaw gives rise to appearance of bulldog facies seen as stigmata of late congenital syphilis.

d.Old man facies[34]: seen in congenital syphilis with marasmus, pot belly, old man facies and withered skin.

Facies of systemic sclerosis[35]:

a.Mask like facies: The facial appearance in a well-developed case is characteristic. The forehead is smooth and shiny. The skin is bound down and hard, the lines of expression and smoothed out and nose becomes small and pinched. The mouth opening is constricted and radial furrows appear giving a pursed appearance. The lower eyelids cannot be depressed by the fingers to show the conjunctiva because of atrophy of tissues. Mat-like telangiectases are frequently found, mandibular atrophy can occur.

b.Mouse like facies: nasal alae become atrophied resulting in pinched appearance of the nose.

- Thyrotoxic facies (Grave's disease): The facie of hyperthyroidism depends chiefly upon "stare". Alert, startled, flushed and anxious appearance. Protrusion of of one or both eyes (exopthamlos) associated with retraction of the upper eyelids (lid lag) which results in the exposure of white conjunctiva above the cornea (Von-Graef's sign). Joffroy's sign- eye movements are often diminished in range and the muscle of brow are wasted, giving diminished wrinkling on raising the eyebrow.
- Torpid or Myxedematous (Myxedema): Skin generally thickened, coarse, dry, pale and waxy with a tinted rose- purple flush over each cheek. tongue is enlarged, broadened nose, ears are thickened, lips swollen, periorbital oedema, xanthelasma, coolness and dryness of skin and hair and alopecia.
- Triangular facies[36]: seen in osteogenesis imperfect, and immunodeficiency, centromeric instability and dysmorphism syndrome. Facial dysmorphism is variable, but common features include low set ears, hypertelorism, flat nasal bridge, epicanthic folds, tongue protrusion and micrognathia.
- Facies in Turner's syndrome[37]: Webbed neck (pterygium colli), low posterior hair line, low misshapen ears, high arched palate, cutis laxa on neck and buttocks.
- Whistling facies: Freeman Sheldon Syndrome is characterized by a typical "whistling" facies [38]consisting of a small, pursed mouth; long philtrum, small nose, deeply sunken eyes and scar like contracture that extends from the middle of the lower lip to the chin.

Conclusion

In the deviations from the 'usual faces' lie subtle clues. These clues may point the keen observer not just to myriad possibilities but can be the tell-tale symptom of the disease itself. In this article we have attempted to compile and classify in one place, the various facies associated primarily with dermatological conditions. This narrative serves to academic interests as well as to the clinician's inquisitiveness. However, the enumeration, while attempting to include as many descriptions as possible, is non-exhaustive. The reader should note that while some facies could be pathognomic for certain conditions, there could be multiple causes for some. The knowledge of disease pathogenesis, processes and more importantly, ways to treat and rehabilitate, add completeness to these interesting presentations in our clinics.

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