Resident's Page

Characteristic facies: An index of the disease

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Reading face is an art. This art has significance in dermatology, a visual specialty. In medical context, facies are distinctive facial expressions associated with specific medical conditions. Here, we have compiled a list of characteristic facies that can aid in diagnosing many diseases and syndromes [Table 1].

Table 1: Characteristic facies in dermatology

Characteristic facies	Description
Acrogeric facies	Acrogeric face is seen in type IV Ehlers–Danlos syndrome. These patients, even at a young age, appear prematurely aged. The face has a pinched appearance with prominent eyes, thin nose, thin lips, and lobeless ears.[1]
Acromegaloid facies	It is characterized by generalized expansion of the skull at the fontanelle, pronounced brow protrusion, often with ocular distension, pronounced lower jaw protrusion with macroglossia and gaping teeth. Such patients also presents with hypertrichosis, hyperpigmentation, hyperhidrosis, acrochordons, enlargement of the hands, feet, nose, lips, ears and a generalized thickening of the skin. It is seen in familial partial lipodistrophy.[2]
Adenoid facies	Adenoid facies is the long, open-mouthed and dumb expression on faces of children with adenoid hypertrophy. The characteristic facial appearance consists of underdeveloped thin nostrils, short upper lip, prominent upper teeth, crowded teeth, narrow upper alveolus, high-arched palate and hypo-plastic maxilla. Adenoid facies can be part of Cowden syndrome.[3]
Amiodarone facies	Administration of amiodarone may cause a phototoxic eruption or a brown or blue-gray discoloration of sun-exposed skin. Biopsy specimens of the latter show yellow-brown granules in dermal macrophages, which represent intralysosomal accumulation of lipids, amiodarone and its metabolites.[4]
Angelic facies	Chronic infantile neurological cutaneous and articular (CINCA) or neonatal onset multisystem inflammatory disease (NOMID) syndrome typically presents as nonpruritic urticarial erythema during the first week of life. Most patients show a typical angelic facies with frontal bossing, a saddleback nose and midfacial hypoplasia.[5]
Antonine facies	In tuberculoid leprosy, involvement of facial nerves produces a blank expression described as antonine facies.[6]
Asiatic porcelain doll facies	It is characterized by tight skin, sparse or absent eyelashes, a fixed facial expression, blurring of groove between nose and cheek, micrognathia, mouth in the "O" position, rigid and tense skin with erosions and denudations and multiple joint contractures. It is seen in restrictive dermopathy.[7]
Atopic facies	In childhood atopic dermatitis, the cheeks are quite pale, there is periocular darkening with dry, slightly scaly skin surface and quite a characteristic facial appearance. The most typical clinical changes in atopic dermatitis are seen around the eyes and together with the pallor and dryness mentioned above gives rise to a characteristic set of features known as atopic facies.[8]
Bird-like facies	A popular term for the facial dysmorphia, characterized by high-arched cleft palate, micrognathia and glossoptosis result in a bird-like face. Bird-like facies may appear alone or in association with Hallermann–Streiff syndrome,[7] familial partial lypodystrophy,[9] Nijmegen breakage syndrome,[10] and Hutchinson–Gilford progeria.[11]

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Table 1: Contd...

Characteristic Facies	Description
Bloodhound facies	It is seen in generalised cutis laxa with lax skin and loss of elasticity.[12]
Bull dog face	It is seen in congenital syphilis as a result of local effect of syphilitic rhinitis on the development of adjacent structures. Because the maxilla is small, the normal mandible appears proportionately longer and bigger resulting in bull dog-like jaw.[13]
Cadaveric facies	Congenital generalized lipodystrophy is characterized by generalized loss or absence of metabolically active subcutaneous fat from birth, resulting in a cadaveric facies and a distinctive muscular body habitus.[2]
Cachectic facies	Human immunodeficiency virus (HIV) patients who are on highly active antiretroviral therapy (HAART) may develop peripheral lipoatrophy with loss of subcutaneous facial fat, particularly buccal, parotid, and preauricular fat pads resulting in prominent zygomata, sunken eyes, deepened and redundant melolabial folds, and a cachectic facies.[2] [Figure 1].
Cigarette facies	Cigarette face is characterized by pale, grey, wrinkled skin with rather gaunt features, so that heavy smokers can often be recognized from their facial appearance alone.[14]
Coarse facies	The typical facial changes include short nose, flat face, and a large head. Their heads tend to be longer than normal from front to back, with a bulging forehead due to premature fusion of skull bones. Individuals with coarse facies look remarkably similar due to coarsening of their facial features. They can be seen in various dermatological conditions like hyper-IgE syndrome,[10] multiple sulphatase deficiency,[15] Costello's syndrome,[14] and galactosialidosis.[16]
Dengue facies	Dengue fever is accompanied by the dengue facies characterized by flushing, palpebral edema, conjunctival injection, retroocular pain, and photophobia. As the symptoms abate, a morbiliform or scarlatiniform rash appears.[17]
Dog/Simian facies	It is seen in congenital hypertrichosis lanuginose. In this condition, most of the body is covered with fine, blond or nonpigmented hair at birth. These hair may reach up to 10 cms and often blend with the darker, terminal hairs of the scalp and/or eyebrows. Whorls of hair may be noted around the sacrum, preauricular region and pinnae.[18]
Drawn facies	This is described in POEMS syndrome, which is an acronym for polyneuropathy, organomegaly, endocrine disorders, M protein and skin changes.[19]
Dysmorphic facies	This is described in patients of DiGeorge syndrome[20] who have low-set, abnormally formed ears, hypertelorism, antimongoloid slant, micrognathia, short philtrum to the upper lip, high-arched palate, palatal abnormalities like cleft palate, and velopharyngeal insufficiency.
Elfin facies	Patients with elfin facies bear facial characteristics similar to that of elves. Because of imprecision inherent in such a definition as well as the potential for offense, it is better to use more specific terminology when possible, such as low-set ears. It can be associated with Williams syndrome.[21]
Facies leprosa	Facies leprosa is characterized by a combination of nasal change and resorption of nasal bone, anterior nasal spine, supra-incisive alveolar region, and anterior alveolar process of the maxillae, associated with the loss of upper incisors teeth, according to the criteria of radiographic interpretation.[22] The clinical aspect of facies leprosa is termed Bergen Syndrome I (nasal leprosy) and Bergen Syndrome II (the leprogenic changes of the alveolar process of the maxilla).
Gargoyles facies	Gargoylism is characterized by thickening and coarsening of facial features due to subcutaneous deposition of mucopolysaccharides.[23] The head is large and dolichocephalic, with frontal bossing and prominent sagittal and metopic sutures, and the face exhibits mid-face hypoplasia, depressed nasal bridge, flared nares, a prominent lower third, thickened facies, widely spaced teeth with attenuated dental enamel, and gingival hyperplasia.
Gaunt facies	The long-term use of highly active antiretroviral drugs (HAART) has been commonly associated with lipodystrophy, producing gaunt facies with sunken cheeks.[24] [Figure 2].
Grimace-like facies	In progressive systemic sclerosis, appearance of the face is characteristic. The forehead is smooth and cannot be wrinkled and atrophy and tightening of the skin gives a characteristic appearance due to fixed stare, pinched nose, prominent teeth, pursed lips, reduced oral aperture and a perpetual grimace-like facies.[25]
Greek helmet facies	Wolf–Hirschhorn syndrome presents with severe growth retardation and mental defect, microcephaly, "Greek helmet" facies, cleft lip or palate, coloboma, cardiac septal defects, aplasia cutis of the scalp and seizures.[26]

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Characteristic Facies	Description
Hippocratic facies	It is characterized by a pinched expression of the face with sunken eyes, relaxed lips, and hollow cheeks and temples, as observed in one dying after an exhausting illness.[27]
Latrodectus facies	The grimacing facial expression seen with Black Widow Spider (*Latrodectus mactans*) bite has been called Latrodectus facies.[28]
Leonine facies	Lion-like face due to diffuse infiltration of the skin of forehead, chin, nose, and ears is seen in various conditions like leprosy [Figure 3], chronic actinic dermatitis, cutis verticis gyrata, leishmaniasis, lymphoma, leukemia, mycosis fungoides, multicentric reticulohistiocytosis, multiple keratoacanthoma syndrome (Grzybowski type), progressive nodular histiocytoma, sarcoidosis, scleromyxedema.[29]
Mask-like face	An immobile, expressionless face with staring eyes and slightly open mouth. It is sometimes associated with Parkinsonism or psychiatric conditions and scleroderma (scleroderma mask) [Figure 4].[30]
Mickey mouse facies	Children with Cockayne's syndrome are short and look cachectic. Weight is affected more than height; hence, it is termed "cachectic dwarfism." Microcephaly and cachectic look give them "Mickey-mouse like" facies.[31]
Mongoloid face/flat facies	In Down's syndrome, the characteristic facial features include flat facial profile, hypertelorism, Mongoloid slant, epicanthal folds, Brushfield spots on the iris, low-set ears, saddle nose, narrow, short, high-arched palate, smaller teeth, and scrotal tongue.[32] [Figure 5]
Monkey facies	Marasmus patients have a wrinkled, loose and dry skin. There is a substantial loss of subcutaneous fat tissue and the facial expression is described as "monkey facies" due to the loss of the buccal adipose tissue.[16]
Moon facies	People with moon facies have round, full or puffy face. The sides the face may become so round from the buildup of fat that ears cannot be seen from the front. Fat deposits in the sides[33] of the skull can also make the face look rounder. It is seen in Kwashiorkor, Cushing's syndrome and patients on steroid treatment [Figure 6].
Old man facies	It is described in congenital syphilis.[33]
Progeroid face	Progeroid type of Ehler Danlos[34] syndrome patients show triangular, old-looking face with a beak shaped or pinched nose, pseudohydrocephalus, wide fontanelles, prominent veins, especially, of the scalp and sparse scalp hair. They also have a distinct pattern of lipodystrophy involving subcutaneous fat loss from the face, extremities, paravertebral and lateral gluteal regions.
Sad man facies	In staphylococcal scalded skin syndrome, erythema often first appears on the head with generalization within 48 hours. Later, the skin develops a wrinkled appearance owing to the formation of flaccid bullae within the superficial epidermis. In 1-2 days, the bullae are sloughed, leaving behind moist skin and areas of thin, varnish-like crust. Patients demonstrate "sad man" facies, perioral crusting, and radial fissuring with mild facial edema.[35]
Slapped cheek facies	In erythema infectiosum, diffuse erythema and edema of the cheeks gives "slapped cheek" facies appearance.[36]
Tabetic facies	It is described in tabes dorsalis. Ptosis and flabbiness of the facial muscles probably contribute in a large measure to the so-called tabetic facies.[37]
Triangular face	In immunodeficiency, centromeric instability, facial dysmorphism (ICF) syndrome,[10] facial dysmorphism is variable, but common features include low-set ears, hypertelorism, flat nasal bridge, epicanthic folds, tongue protrusion and micrognathia. This is described as triangular facies. Triangular facies has also been described in Ehler–Danlos Syndrome[38] and osteogenesis imperfecta.[39]
Whistling facies	Seen in Freman–Sheldon syndrome that shows characteristic whistling face with craniocarpotarsal dysplasia and skin dimpling defect.[40]

Beside these there are various conditions that have characteristic facies without specific names, including Noonan's syndrome, Langer–Giedion syndrome, Proteus syndrome, Leprechaunism, Moore–Federman syndrome, Gorlin's syndrome, Hyperpituitarism, Alagille's syndrome

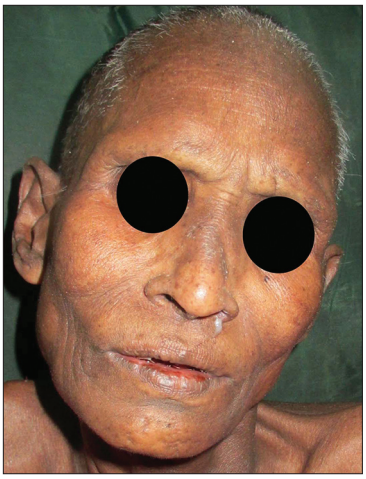

Figure 1: Cachetic facies of human immunodeficiency virus infection

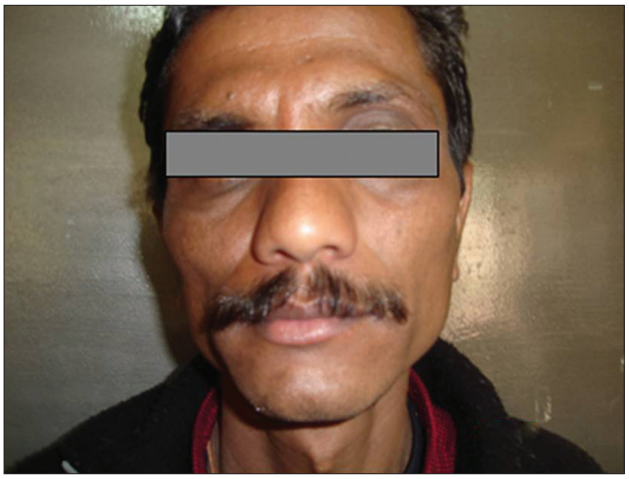

Figure 2: HAART induced gaunt facies with sunken cheeks

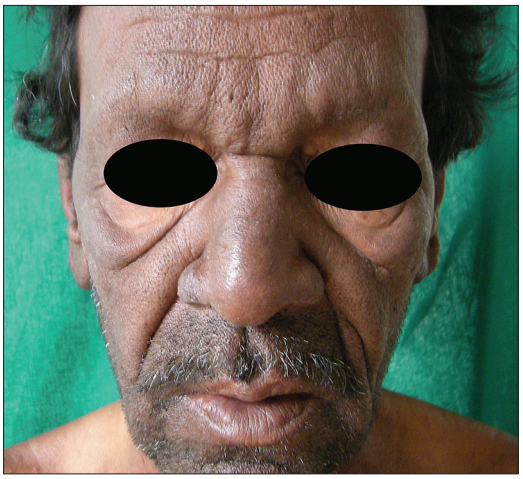

Figure 3: Leonine facies of lepromatous leprosy

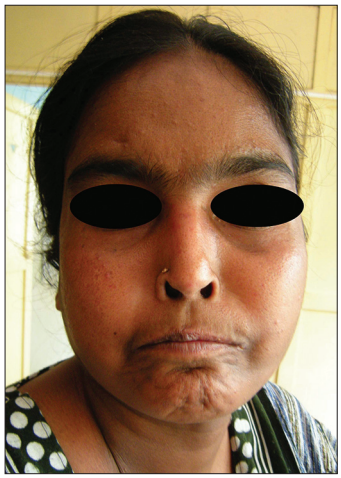

Figure 4: Mask-like facies of scleroderma

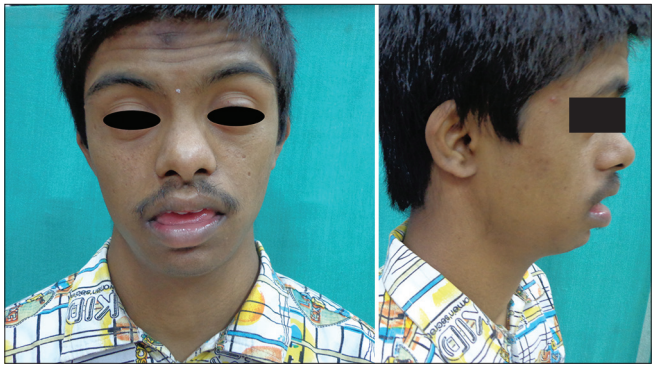

Figure 5: Mongoloid/flat facies of Down's syndrome

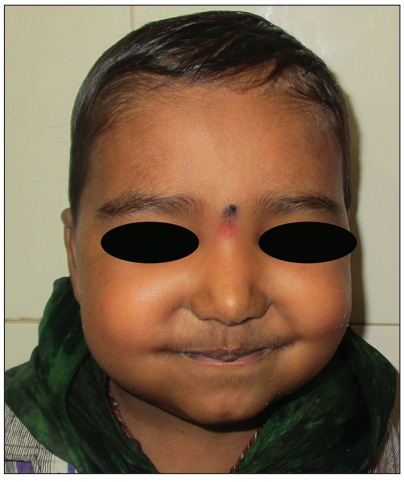

Figure 6: Steroid-induced moon facies in a child of nephrotic syndrome

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