

A SUPPLEMENT TO THE CORE
TUTORIALS IN DERMATOLOGY
FOR PRIMARY CARE

CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

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CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY A SUPPLEMENT TO THE CORE TUTORIALS IN DERMATOLOGY FOR PRIMARY CARE

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The full set of Core Tutorials in Dermatology for Primary Care comprises a prequel on diagnosing skin conditions, a sequel on Tests and Techniques, Which, When and Why? along with chapters on The Eczemas, Psoriasis, Skin Infection and Infestation, Skin Malignancy, Leg Ulcers, Acne and Urticaria and Related Allergic Disorders. These chapters and their accompanying Self Test Questionnaires are available from Dermal Laboratories. They can be downloaded from the Healthcare Professionals Resources section of the Dermal website www.dermal.co.uk or can be requested directly from Dermal at the contact details below.

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INTRODUCTION

The cutaneous manifestations of systemic disease are both fascinating and diverse. Most importantly they can provide the first clues to significant underlying diseases be they metabolic, infective or neoplastic. In this tutorial we will cover the most common of these plus a few rarer, but very distinctive presentations. The sheer diversity of these conditions does not allow for ease of classification, so I have listed them here in alphabetical order.

ACANTHOSIS NIGRICANS

This condition is characterised by asymptomatic pigmentary hyperkeratosis of the flexures, perineum and skin folds of the neck with papillomatosis change in a typical 'velvety' morphology. Classifications include:

- hereditary type (a pattern associated with insulin resistance/diabetes)
- the commonly occurring obesity-related pattern (benign acquired acanthosis nigricans)

There is a rare drug-induced pattern and most importantly a pattern recognised as a paraneoplastic phenomenon, most commonly with adenocarcinomas and more rarely with lymphoma. Such presentations are uncommon and usually manifest with more widespread involvement often with mucosal pigmentary change in addition. Involvement of the palmar aspects of the hands is almost exclusively associated with internal malignancy, particularly gastric carcinoma.

Treatment relates to identifying the underlying cause. For the benign acquired variety, a significant weight loss can produce resolution in up to 50% of cases. For the paraneoplastic associated variety, it is important to note this phenomenon can significantly pre-date the diagnosis of malignancy. Treatment of the underlying tumour may result in disease regression.

ACANTHOSIS NIGRICANS



ACANTHOSIS NIGRICANS





CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

AMYLOIDOSIS

Amyloid can be deposited abnormally in a variety of tissues and has a wide range of disease associations. There are both acquired and hereditary patterns. It can be a primarily localised cutaneous disease with various sub-types, most commonly the macular and papular varieties which may co-exist and there is also a rarer tumescent variation. Primary skin amyloid is of unknown aetiology. The papular form is the most commonly recognised, presenting as a chronic, itchy eruption with a predilection for the shins and less commonly, thighs and upper limbs. Differential diagnosis would include lichen planus and lichen simplex chronicus.

Macular amyloid can often present clinically very subtly and can be easily mistaken for post-inflammatory hyperpigmentation. The chest and back are the most commonly involved sites. It can be completely asymptomatic. Histology is characteristic, but treatment options are limited and disappointing.

CUTANEOUS AMYLOIDOSIS



'RACoon SIGN' OF PERIORBITAL AMYLOID DEPOSITION



CROHN'S DISEASE

CROHN'S CHEILITIS



There are a number of cutaneous manifestations associated with Crohn's disease. These characteristically manifest at each end of the gastro-intestinal tract, either as a granulomatous cheilitis of the lips or fissuring erosion and ulceration of the peri-anal and perineal skin.

PERI-ANAL CROHN'S DISEASE



Up to 25% of cases will manifest some perineal involvement. Peri-stomal tissue can also be involved. Crohn's disease can also involve distant sites, so-called 'metastatic' Crohn's, where it can manifest as ulcers, nodules, abscesses, plaques or pustules.

Pyoderma gangrenosum, (discussed in greater detail on page 12) has an association with both Crohn's disease and ulcerative colitis. Disease activity does not correlate consistently with the severity of internal disease.



CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

DERMATITIS HERPETIFORMIS

Noted to be one of the most intensely pruritic of all skin conditions. There is an association with gluten-sensitive enteropathy/coeliac disease. DH is an autoimmune blistering condition mediated via IgA which runs a chronic course. Characteristically the extensor aspects of knees, elbows and the buttock and scalp are involved. Vesicles may not be clinically apparent as they have often been ruptured by excoriation. The lesions heal without scarring.

DERMATITIS HERPETIFORMIS



The incidence range is 0.4 – 2.6 per 100,000 in Northern Europe and onset is typically in the fourth decade with a slight preponderance in men. The severity of co-existent gluten sensitivity is extremely variable but all patients with this clinical diagnosis require investigation by a Gastroenterologist. There is also an increased risk of small bowel lymphoma. Diagnosis is established by skin biopsy and direct immunofluorescence. The mainstays of treatment are a gluten-free diet and the option of adding in oral dapsone.

DERMATOMYOSITIS

This constitutes an autoimmune disorder of both skin and skeletal muscle.

There is a wide spectrum of disease. In some cases, the myopathy dominates, in others the cutaneous involvement. Although rare, it is important to be aware of this condition, not least because of its strong association with internal malignancy and indeed it often constitutes the first presentation of such. Rarer again, is the juvenile form. The clinical presentation is identical but there is no association with malignancy in the paediatric age group. The majority of malignancies are adenocarcinomas, particularly of the breast, ovary and lung. 50% of patients present with the characteristic skin rash, most commonly on the face with peri-orbital heliotrope discolouration. There may also be oedema and alopecia.

DERMATOMYOSITIS – PERI-ORBITAL HELIOTROPE DISCOLOURATION



Another characteristic cutaneous presentation is a linear rash involving the extensor aspects of the digits along with Gottron papules on the interphalangeal joints and extensor surfaces of the elbows and knees. Often there is also a rash involving the upper chest. Prominent nail fold telangiectasia and ragged cuticles can also be manifest.

There is an associated proximal myositis presenting as weakness and fatigue. This can be quite insidious in onset but can become severe and progressive. Involved skin may occasionally erode and ulcerate.

Investigation revolves around creatine kinase levels and antibody assays along with histology, both of muscle and skin to exclude an underlying malignant process. It can be variable, and prognosis relates to any underlying pathologies. Most patients require extended courses of immunosuppressive medication.

DERMATOMYOSITIS – GOTTRON PAPULES



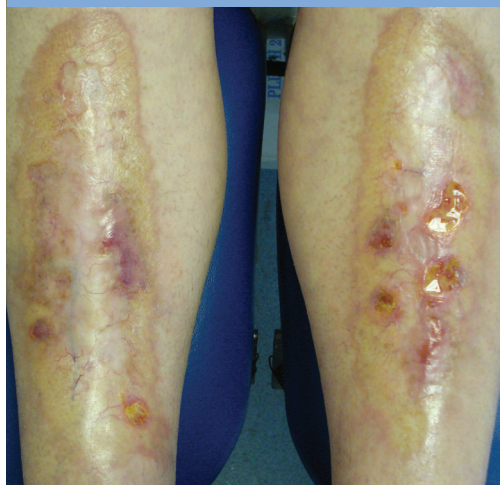


CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

DIABETES MELLITUS

There is quite a range of diabetic involvements of the skin including a general increased risk of a wide range of infections, diabetic ulcers and injection site reactions. The two, we will discuss in more detail here are necrobiosis lipoidica (diabeticorum) and diabetic bullae.

NECROBIOSIS LIPOIDICA



DIABETIC BULLAE



Necrobiosis lipoidica has strong associations with both diabetes and impaired glucose tolerance albeit perhaps not so strongly as previously thought. It can occur in between 2 and 3% of the type 1 diabetic population. These are distinctive with the development of well-defined erythematous plaques most commonly over the pre-tibia area. These have characteristic yellowish atrophic centres. They can progress to ulceration and be extremely challenging to treat/heal. Young adults are the most likely affected group.

Bullae: These present as non-scarring large sub-epidermal bullae most commonly on the lower limbs. Although not common there is a strong correlation with type 1 diabetes. They self-resolve over a number of weeks.

ERUPTIVE XANTHOMATA

These manifest as multiple firm yellowish papules on the extensor surfaces of elbows, knees, back and buttocks measuring 2 – 5 mm. They are usually asymptomatic. Their clinical significance is the association with underlying disorders of lipid metabolism. A full lipid profile with emphasis on triglyceride levels needs to be assessed. The lesions themselves resolve with normalisation of the lipid profile following treatment.

ERUPTIVE XANTHOMATA



ERYTHEMA NODOSUM

This clinically presents with the acute development of symmetrical, tender, erythematous subcutaneous nodules measuring 1 – 5 cm and most commonly in the pre-tibial areas. They characteristically ulcerate; recurrent episodes are common and represent a non-specific reactive process triggered by a wide range of disparate conditions. The most well-established triggers are:

- drugs, particularly the oral contraceptive pill
- infections such as streptococcal and mycobacterial
- sarcoidosis
- inflammatory and infective bowel disease

ERYTHEMA NODOSUM



Prevalence is high at 2.4 per 1000 per year in the 20 – 40 age group with a much higher incidence in females.

Investigations are determined by careful history taking but might include full blood count, inflammatory markers, antistreptolysin O titre (ASO) titres, urinalysis, throat swab, chest x-ray and tuberculosis (TB) screening. Histology demonstrates a septal panniculitis without vasculitis. Biopsies are required for examination. Non-steroidal anti-inflammatories are first-line treatment unless contra-indicated (e.g. inflammatory bowel disease).

LIVEDO RETICULARIS

This is a descriptive term for a pattern of cutaneous involvement comprising a mottled cyanotic discolouration with a reticulated mesh, or lace-like pattern beneath the skin. The appearances are caused by abnormalities in the underlying vascular network, often accentuated by cold.

IDIOPATHIC LIVEDO RETICULARIS





CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

Most cases are physiological or idiopathic. Livedo reticularis (cutis marmorata) occurs in 50% of children and is purely a vascular reaction to cold. It is most commonly manifested in the limbs. Pigmentary change or superficial scaling can indicate external thermal damage (erythema ab igne) most commonly seen in the lower limbs of elderly patients' consequent on sitting in too close proximity to the fire and classically after hot water bottle use.

There is also an acquired idiopathic pattern of livedo with a significant association with migraine variant with systemic involvement (Sneddon Syndrome) which demonstrates progressive livedo with arterial change, essentially equating to an endarteritis which may clinically manifest as migraine/stroke in young adult women. Such patients have positive cardiolipin, anticardiolipin and anti-phospholipid antibodies and a third will have a co-existent diagnosis of systemic lupus. Secondary livedo reticularis has many causes including a range of connective tissue and autoimmune diseases. It can also be a result from infection, drug reactions and be a paraneoplastic phenomenon.

CUTANEOUS MASTOCYTOSIS

(AKA URTICARIA PIGMENTOSA)

A rare acquired condition resulting from an excess of mast cells in the skin and other tissues with a prevalence estimated at 1 in 10 to 30,000 of population. It can manifest in children from birth onwards; in the paediatric population it is usually self-limiting and resolves. If it occurs after the age of 10 it is usually persistent and there can be associations with systemic disease. Patients can present with a single mastocytoma or a symmetrical eruption composed of well-demarcated red-brown elevated macules, papules, plaques or nodules approximately 1 cm in diameter with sparing of the palms and toes. This may be vesicular in young children. Mucous membrane involvement is recognised. Symptoms are very variable, but itch can be significant. Supported by eliciting a positive Darier's sign, that is the lesions will urticariate if rubbed. Very occasionally there may be systemic involvement with internal mastocytosis especially of the bones. Such lesions can be recognised radiologically. Systemic treatment can be monitored through haematological markers. There can be both gastrointestinal (GI) and reticular endothelial involvement. Malignant transformation is a rare complication. Diagnosis can be confirmed by biopsy, during this procedure local anaesthetic needs to be injected peripherally to avoid mast cell degranulation that can lead to diagnostic uncertainty.

Investigation for systemic involvement includes assay of serum tryptase levels and bone marrow biopsy. Treatment centres around the use of histamine antagonists – H1 and H2 blockers with phototherapy reserved for severe or resistant cases.

CUTANEOUS MASTOCYTOSIS



CUTANEOUS METASTATIC DISEASE

Although much less common than hepatic spread many malignancies can metastasise to the skin and indeed this can be the initial clinical presentation to static cancer. Of patients with metastatic disease, up to 10% will develop cutaneous metastases. Most frequent are breast, lung, colon, upper GI tract, uterus and kidney. In my own clinical dermatological experience, I have seen renal, pancreatic and breast secondaries along with haematological and lymphoproliferative diseases such as leukaemia and lymphoma. The development of firm, painless nodules is the most common clinical presentation and if such is the case, one of the golden rules of dermatology must always be followed, that is that all EFG (evaluated, fixed and growing) lesions require histology. The most common skin metastases from previous unidentified tumours are kidney, lung, thyroid and ovary in origin.

LYMPHOMA



SKIN METASTASIS FROM RENAL CARCINOMA



ADENOCARCINOMA OF LUNG



CUTANEOUS LEUKAEMIA





CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

NEUROFIBROMATOSIS

This is a genetically inherited neuro-cutaneous disorder. There is a sub-division between two distinctive types: NF1 and NF2 but there also exists a number of variants. NF1 is by far the most common comprising 80% of all cases at a prevalence of 1 per 3000 - 5000. The incidence pattern is autosomal dominant. Spontaneous mutations account for approximately half of all cases.

Dermatological signs are:

A: Café au lait macules

These can be evident at birth or develop during the first year of life and increase in size and number over the first decade and the existence of 6 or more such macules are significant.

B: Flexural freckling

This also develops in early life most commonly in the axillae and is evident in the majority of cases.

C: Neurofibromas

These characteristically do not appear until puberty and increase in incidence with age. The trunk is the most common site. They can present with either a pedunculated or polymorphic morphology and can be rarely segmental in distribution. One in 10 patients exhibit plexiform neuromas which track along nerves and are characteristically tender. More severe forms can cause both bony and soft tissue deformity. The risk of malignant progression is approximately 15%, so rapid change in size or increasing tenderness in a lesion needs careful evaluation.

There is also an increased risk of other neurological tumours as well as malignancy in other tissues particularly optic gliomas. Early onset and rapid progression of cutaneous lesions equate to a poor prognosis. Lisch nodules can be detected on ophthalmic examination.

Management requires a multidisciplinary approach and is largely symptomatic.

NEUROFIBROMATOSIS – CAFÉ AU LAIT MACULES



OSLER-WEBER-RENDU DISEASE

(AKA HEREDITARY HAEMORRHAGIC TELANGIECTASIA)

This is a condition that can be inherited in an autosomal dominant pattern affecting 1 in 50,000, with 20% of cases arising sporadically. The cutaneous manifestations are of multiple telangiectasia of the lips/ oral cavity and skin. Children often present with frequent epistaxis or complications can occur in adults with bleeding from involvement in the GI tract.

OSLER-WEBER-RENDU DISEASE



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PEUTZ JEGHERS SYNDROME

This is an autosomal dominant mediated condition that manifests with multiple oral and peri-oral lentiginos. 40% are new mutations. The important association is with polyposis of the GI tract with a risk of malignant transformation and affected patients require a regular screening programme.

PEUTZ JEGHERS SYNDROME



PORPHYRIA CUTANEA TARDA

The porphyrias are a rare and most commonly inherited group of metabolic disorders resulting in toxic levels of porphyrins. The one most commonly encountered in the UK is porphyria cutanea tarda which can be both inherited and acquired. This has a strong association with alcohol excess with disorders of iron metabolism specifically of haem biosynthesis and other liver pathologies, such as hepatitis. There is also an association with haemodialysis. This type of porphyria manifests only in the skin, presenting with fragile blisters and erosions typically on the dorsal aspect of the hands and fingers healing to leave characteristic milia. Other clinical signs may include scarring alopecia, hyperpigmentation and hirsutism.

PORPHYRIA CUTANEOUS TARDA



Investigation is by a combination of urine and plasma porphyrin screening and other tests as indicated to investigate possible underlying causes. If a porphyria screen is normal, the possibility of pseudo-porphyrin secondary to drugs, most commonly naproxen, should be considered.



CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

PRURITUS/PRURIGO

Generalised pruritus is a common presentation particularly in the elderly demograph. There is often extensive artefactual damage to the skin and great care must be made not to miss an underlying primary skin condition particularly infestation. A very characteristic clinical finding is the 'butterfly' sign as illustrated here.

BUTTERFLY SIGN



There is a rash consistent with widespread excoriation but with distinct areas of sparing on upper and central back which is perceived to be in the shape of a butterfly denoting the areas that cannot be easily reached by the patient to scratch. This sign, however, cannot be relied on as patients can be quite resourceful to reach any area of skin to scratch it. However, if present it firmly points to the diagnosis not being one of a primary skin condition as the aetiology of itch. Such patients require investigation to identify underlying causes with emphasis on drug history and co-morbidities. An initial blood screen should include urea and electrolytes (U&E's), thyroid function tests (TFTs), full blood count, liver function tests (LFTs), ferritin and inflammatory markers as a minimum and other tests as directed by history and further examination.

PYODERMA GANGRENOSUM

This is a relatively uncommon non-infectious neutrophilic dermatosis with a strong association with underlying chronic inflammatory disease especially bowel related, or a neoplastic process most commonly of the haematological or lymphoreticular systems when an underlying disease or ulcerative colitis is the association in half of all cases. However, 1 in 2 of all cases of pyoderma gangrenosum appear to be idiopathic. It is most commonly of a rapidly evolving erosive necrotising ulceration usually less than 10 cm in diameter. Such ulcers are irregular with a violaceous border and a characteristically undermined and overhanging edges commonly sited on the lower limbs and occurs often with characteristic cribriform scarring. A third of cases are recurrent and systemic upset is variable. The diagnosis is essentially clinical. Histology can be supportive but not distinctive and could be taken to exclude other aetiologies. Treatment centres on the use of immunosuppressant drugs such as prednisolone and ciclosporin. Twenty per cent of cases demonstrate Koebner type phenomenon on an area of previous skin trauma.

PYODERMA GANGRENOSUM



SARCOIDOSIS

Sarcoidosis is a poorly understood antigen mediated condition characterised by the development of non-caseating tubercles in a variety of organs and tissues which can resolve with or without fibrosis over time. Presentation is varied, and any organ can be affected with a predilection for skin, lungs, lymphatics and salivary glands. Age presentation is most commonly between the ages of 20 and 40. The histological features are consistent irrespective of the tissue involved.

SARCOIDOSIS



SARCOIDOSIS OF THE ARMS





CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

Symptoms are non-specific and organ dependant. Approximately 25% of patients with systemic disease will demonstrate dermatological manifestations. The condition can demonstrate the Koebner phenomenon (selective involvement of scarred or traumatised tissue). Cutaneous patterns are varied with papulonodular, subcutaneous, annular and plaque varieties all recognised. The colour is often violaceous. A differential diagnosis clinically would include lupus and cutaneous tuberculosis. Lupus pernio describes a pattern of infiltration of the facial tissues especially around the nose and is more common in older patients. The treatment of cutaneous involvement revolves around the use of oral or intra-lesional steroids. Other therapies would be dictated by the organs involved and the extent of disease.

SYSTEMIC LUPUS ERYTHEMATOSUS

This is a multisystem autoimmune disorder with a predilection for skin, joints, renal and vascular involvement. This is a global disease although relatively uncommon in the UK (6 per 100,000 / year) with a threefold increase in those of Afro-Caribbean origin with a peak age of onset of about 40. There is an increased familial incidence. No specific diagnostic test exists but there are a number of haematological and histological abnormalities which taken together with the clinical features make the diagnosis.

As with the cutaneous variant discoid lupus erythematosus, UV light can be a precipitating or exacerbating factor. A diverse range of drugs and infections can also constitute exogenous triggers. There is a range of cutaneous manifestations including vasculitis, bullae formation, erythema multiforme type lesions, annular erythemas and scarring alopecia. The most striking of the clinical presentations are of the bilateral malar 'butterfly' rash presentations are protean, and the skin may be completely uninvolved in the minority of patients. Treatment requires a multidisciplinary approach. Some patients can have a significantly shortened life expectancy especially in the presence of renal or vascular involvement.

SYSTEMIC LUPUS ERYTHEMATOSUS



SUB-ACUTE CUTANEOUS LUPUS ERYTHEMATOSUS



SUBCORNEAL PUSTULAR DERMATOSIS (AKA SNEDDON-WILKINSON DISEASE)

Although rare this condition has a very characteristic clinical presentation and an association with a number of significant pathologies and is therefore worthy of inclusion in this article. It most commonly presents in the 40 – 60 age group with a female to male ratio of 4 to 1. The condition presents with acute development of areas of pea-sized pustular lesions on an erythematous background and characteristic fluid level of pus, that rupture and coalesce to form an annular or serpiginous pattern and can develop in successive waves.

SUBCORNEAL PUSTULAR DERMATOSIS – ANNULAR, SERPIGINOUS PATTERN



Favoured body sites are the truncal flexures and proximal limbs with facial sparing. A differential diagnosis would include impetigo, pustular psoriasis and dermatitis herpetiformis. Histology is very helpful in differentiating this from other conditions of similar morphology. Fresh intact blisters are optimal for histological examination. The condition commonly follows a benign but chronic course over several years. Important associations are multiple myeloma and connective tissue diseases.



CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

SUBCORNEAL PUSTULAR DERMATOSIS



This syndrome has four significant features:

- acute onset of non-itchy, tender, juicy, well demarcated plaque-like infiltrates
- a fever of greater than 38 degrees centigrade and general malaise
- leucocytosis
- histologically the presence of an intense dermal neutrophilic infiltrate without features of vasculitis

SWEET'S DISEASE

(AKA ACUTE FEBRILE NEUTROPHILIC DERMATOSIS)

SWEET'S DISEASE



Vesicles, bullae or pustules can feature but fever and neutrophilia are not consistent findings. The presentation is essentially reactive and is most commonly manifest in adult females. Provoking factors are underlying malignancy (10 – 20%), particularly haematological, drug reactions, pregnancy and diseases such as inflammatory bowel disease. Many cases are idiopathic. An underlying cause is identifiable in 50% of presentations so all patients require careful clinical evaluation and investigation. The skin changes favour the head and neck and limbs. Eye involvement and mucosal ulcers are common. A differential diagnosis would include erythema multiforme, erythema nodosum and drug reactions. First-line treatment is high-dose systemic steroids.

SYPHILIS

Syphilis is an infectious disease mediated by the spirochete *Treponema pallidum*. Syphilis presents as a cutaneous disease both in its primary and secondary phases. Incidence in the UK population is presently rising albeit from a low base. It is important that there remains awareness of this in the wider medical community as the condition could present to a diverse range of medical professionals and missing a diagnosis at these stages when it is eminently and easily treated can have serious and potential fatal consequences. One diagnostic conundrum is the potential long periods of latency of the disease. The primary chancre occurs at the initial entry point of the organism with a characteristic indurated ulcerated painless papule measuring 1 – 2 mm associated with regional lymphadenopathy. This is usually on the external genitalia or perianal area and self-resolves in a period of normally 4-8 weeks and rarely in excess of 3 months.

Secondary syphilis exhibits a macular papular eruption with 3 common features; an absence of itch, symmetrical distribution and coppery red discolouration. It is often confused with psoriasis and drug eruptions. The timing of onset is approximately 8 weeks after exposure. There may be flu-like constitutional symptoms and generalised lymphadenopathy. Body sites most commonly involved are the angles of the mouth and nose, palms and soles, mucous membranes and flexural and intertriginous areas. Some lesions may be hyperkeratotic. Patchy alopecia may also be a feature. The lesions resolve variably over the course of weeks or months and if the disease goes unrecognised can enter a latent stage with an absence of clinical stigmata. The diagnosis is established by a variety of serological tests that are available.

SECONDARY SYPHILIS





CUTANEOUS MANIFESTATIONS OF SYSTEMIC DISEASES IN DERMATOLOGY

TUBEROUS SCLEROSIS COMPLEX

(AKA BOURNVILLE'S DISEASE, EPILOIA OR ADENOMA SEBACEUM)

This is a genetically determined condition demonstrating an autosomal dominant prevalence varying from 1 in 600 to 1 in 20,000 making the condition one of the most common single gene disorders. The pathogenesis is of a hamartoma formation in multiple organ systems. Sites of predilection are the skin, kidneys, myocardium, eye and brain. In excess of 50% are new mutations. The diagnosis of mental retardation and the co-existence of epilepsy are very variable. The term "Epiloia" historically described a triad of low intelligence and epilepsy and skin adenoma sebaceum.

The characteristic dermatological signs are of hypopigmented 'ash leaf' macules which present in between 40 and 90% of all cases. A Wood's light can be helpful in their identification, also characteristic is peri-ungual fibromata and facial adenomas which both present in up to 50% of cases. The so-called adenoma sebaceum lesions are in fact histologically angio-fibromata. Other characteristic dermatological manifestations are Shagreen patches present in 80% of cases, pathognomonic plaques 1 – 10 cm in size which may be single or multiple and histologically are simply composed of excess collagen. Two thirds of cases manifest dermatological abnormalities. The condition is highly variable in expression and prognosis.

TUBEROUS SCLEROSIS – SHAGREEN PATCHES



VASCULITIS

This is a relatively common non-specific presentation. It can understandably often provoke concern among GPs and requires a pragmatic approach to both diagnosis and treatment. The major players aetiologically are infections in the broadest sense, drug reactions and less commonly, autoimmune and connective tissue diseases. However, in 50% of cases an underlying cause cannot be identified. A vasculitic screen might include general bloods, inflammatory markers, repeated urinalysis profile. Blood pressure needs to be monitored.

The clinical patterns are varied and include erythema, urticaria, purpura, necrosis and infarction. Some distinct clinical patterns should not be missed such as Henoch Schonlein Purpura and in the paediatric population, meningococcal septicaemia and Sweet's disease. Histology can be very helpful. Vasculitis is often mild and self-limiting and often, with appropriate investigation, if the patient remains well, an empirical approach to treatment or a 'watchful waiting' philosophy often suffice.

VASCULITIS



