10e Post O.N.S. Meeting

Stichting Oncowijs

Facial cutaneous physical signs

het gelaat als reflexie van een onderliggende interne maligniteit

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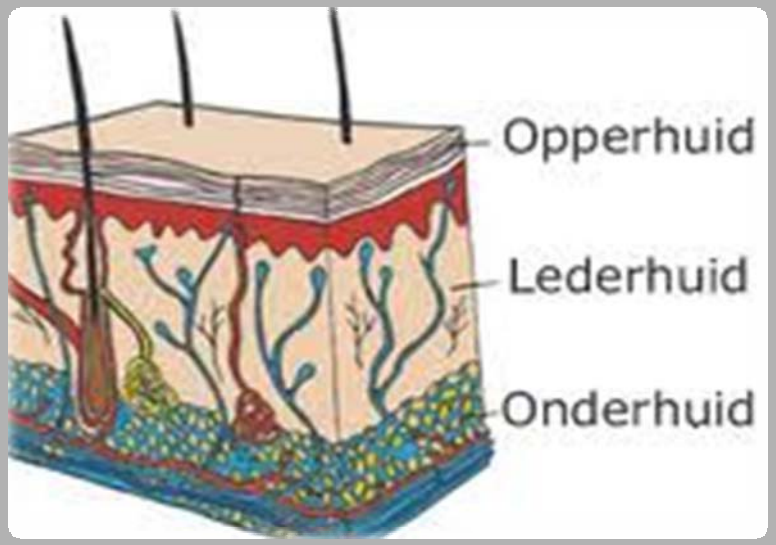
Elkerliek ziekenhuis Helmond

Inhoud van de presentatie

- Huidfeitjes
- Lentiginosis gerelateerde aandoeningen
- Photosensitiviteits gerelateerde aandoeningen
- Hamartoma gerelateerde aandoeningen
- Overige aandoeningen

Huidfeitjes

- Grootste orgaan
- 2 m^2
- 15% van gewicht
- Zintuigelijke functie
- Beschermende functie
- Uniek



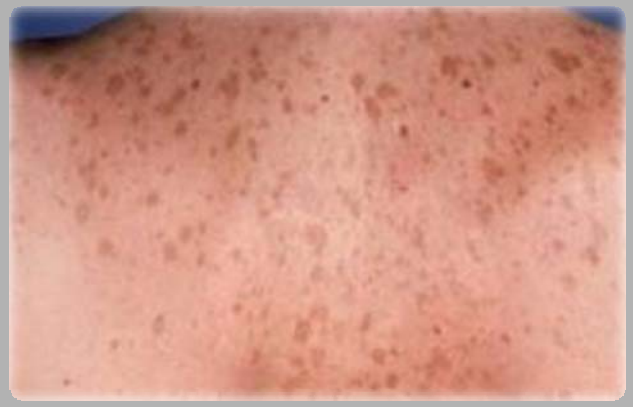

"ALL IS VANITY"

Lentiginosis

- Autosomaal dominant erfelijk
- Mutatie mTOR signaalpad
- Scherp begrensde macula
- Hyperplasie van de melanocyten
- Screening noodzakelijk

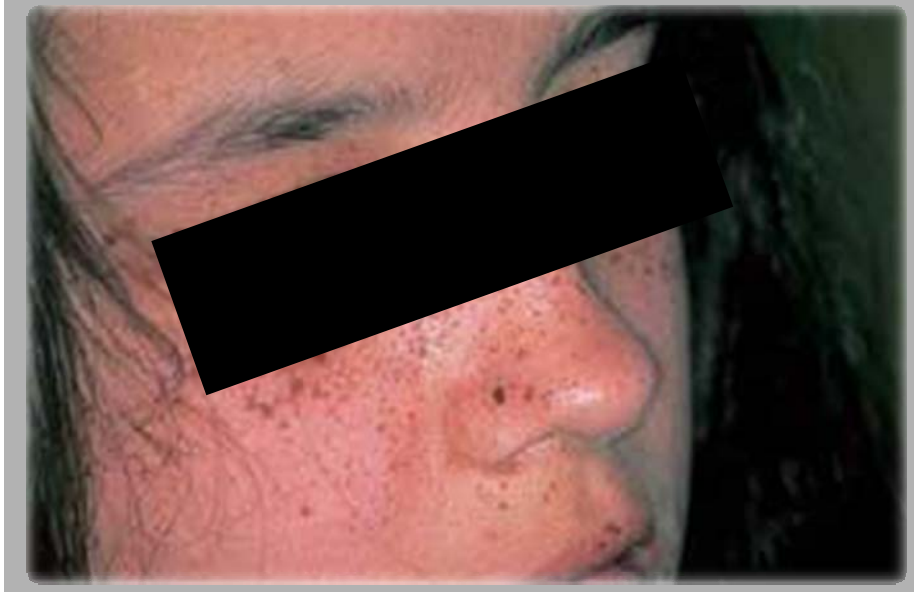
Voorbeelden:

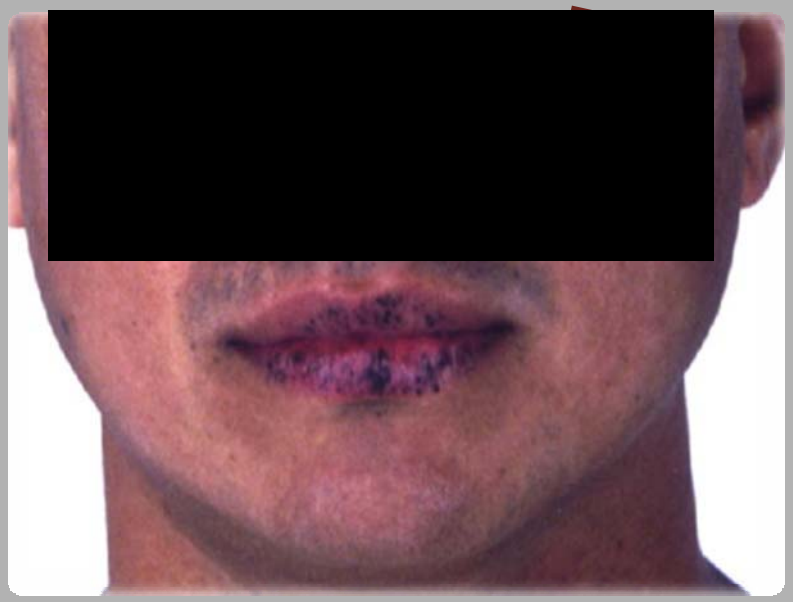
- Peutz-Jeghers syndroom
- Carney complex

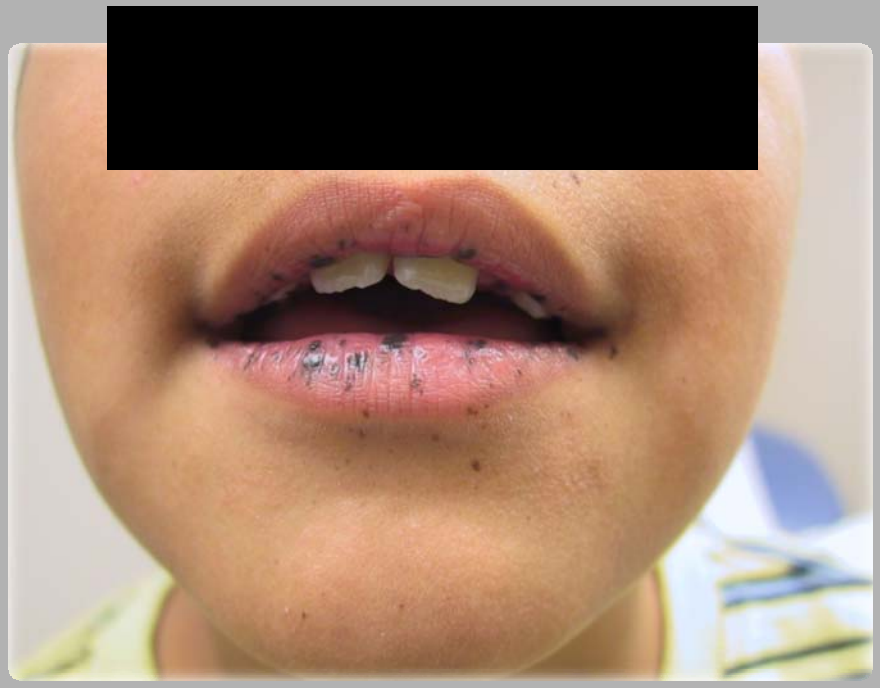


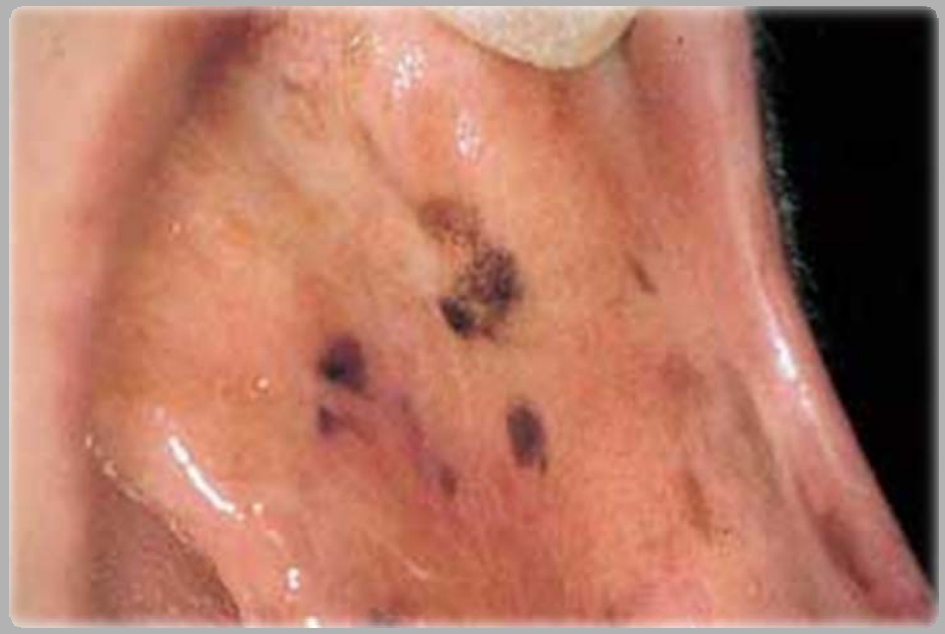
Peutz-Jeghers syndroom

Huidafwijkingen









Peutz-Jeghers syndroom

Systemische afwijkingen

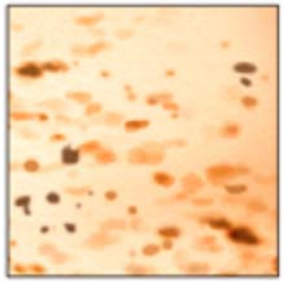
Maligniteiten in:

- Dunne darm
- Uterus
- Ovaria
- Mamma
- Longen
- Testis

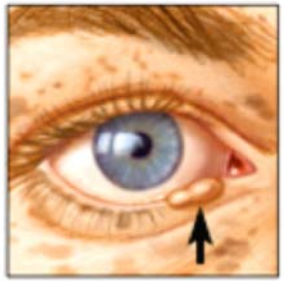
Carney complex

- Zeer zeldzaam
- Gerelateerd aan MEN-syndroom

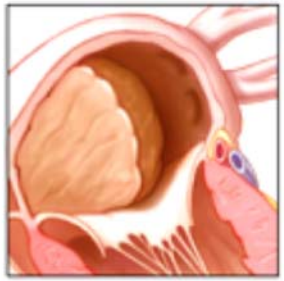
- Huidafwijkingen
- Systemische afwijkingen



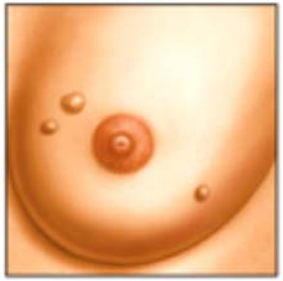
Spotty skin pigmentation, 65%



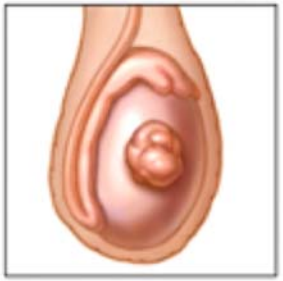
Cutaneous myxomas, 45%



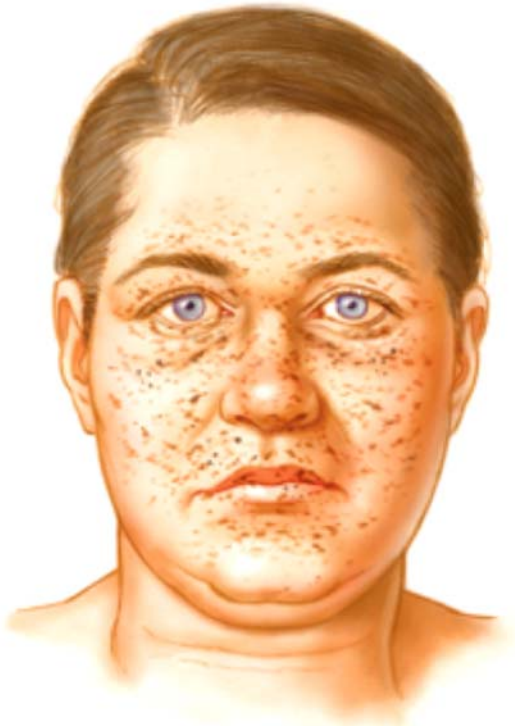
Cardiac myxomas, 72%



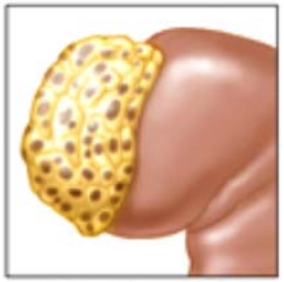
Mammary myxomas, 42%



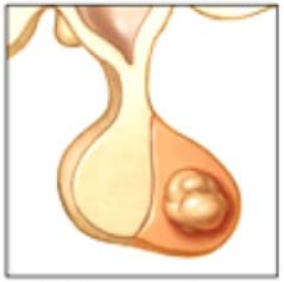
Testicular tumors, 56%



CARNEY COMPLEX

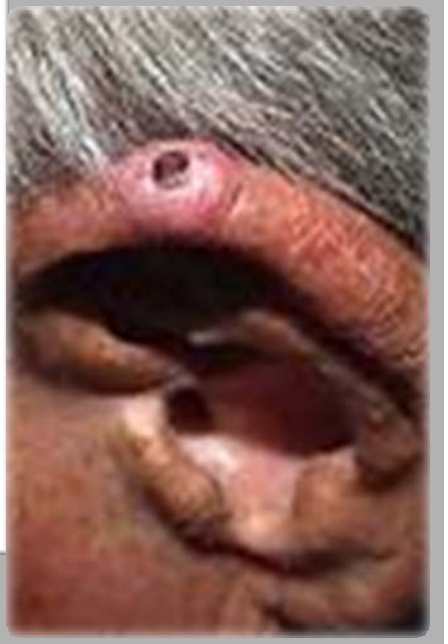


PPNAD, 45%



GH-secreting pituitary tumor, 10%

Schwannomas, 5%



Photosensitiviteits gerelateerde aandoeningen

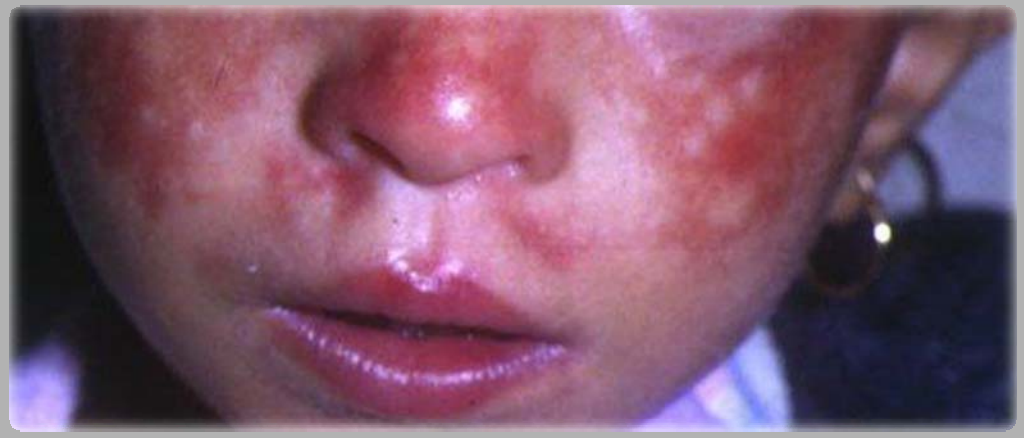
- Genetische afwijking
- Zeldzaam
- Aan (zon)licht blootgestelde huid
- Persisterend en jeukende laesies
- Littekens of huidatrofie

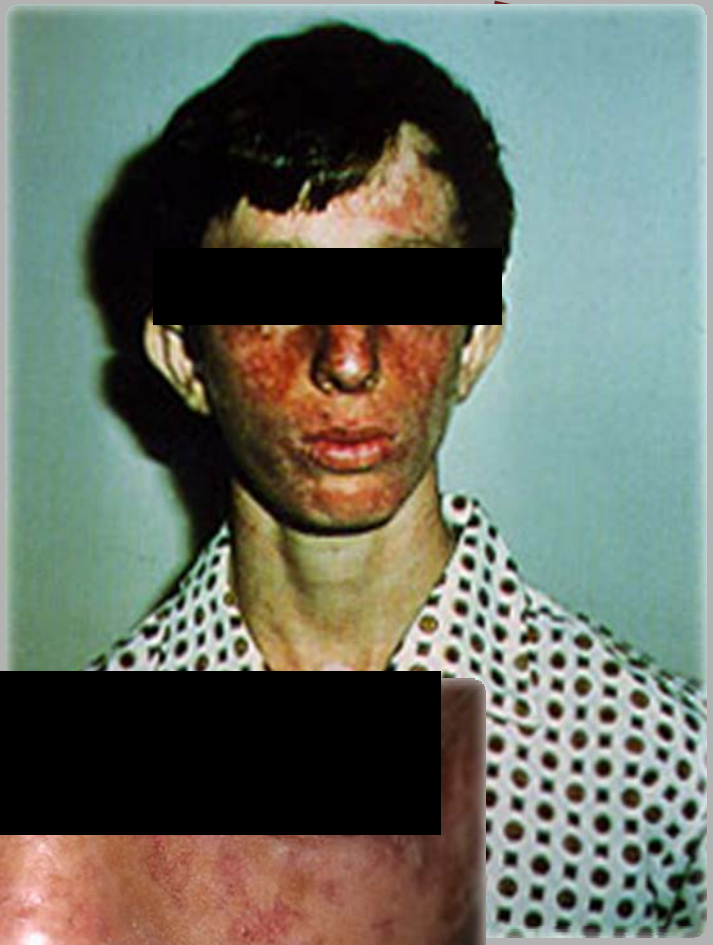
Voorbeelden:

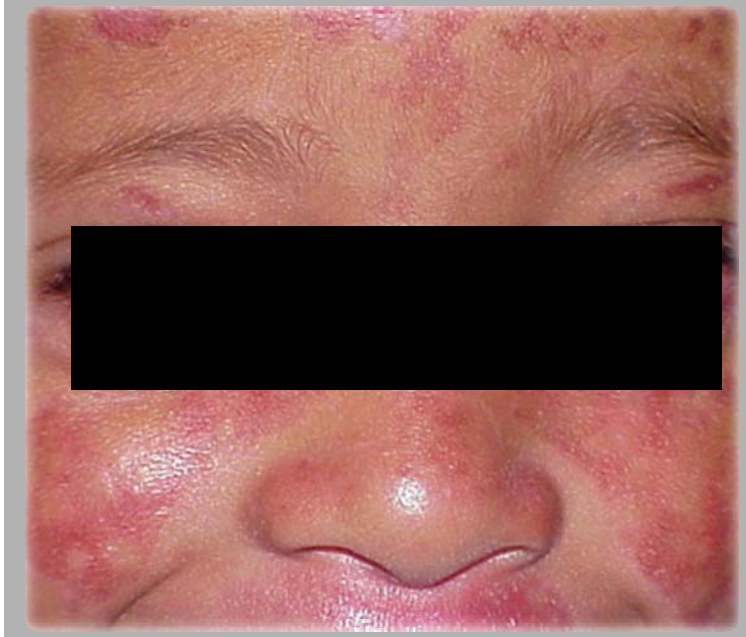
- Bloom syndroom
- Rothmund-Thomson syndroom

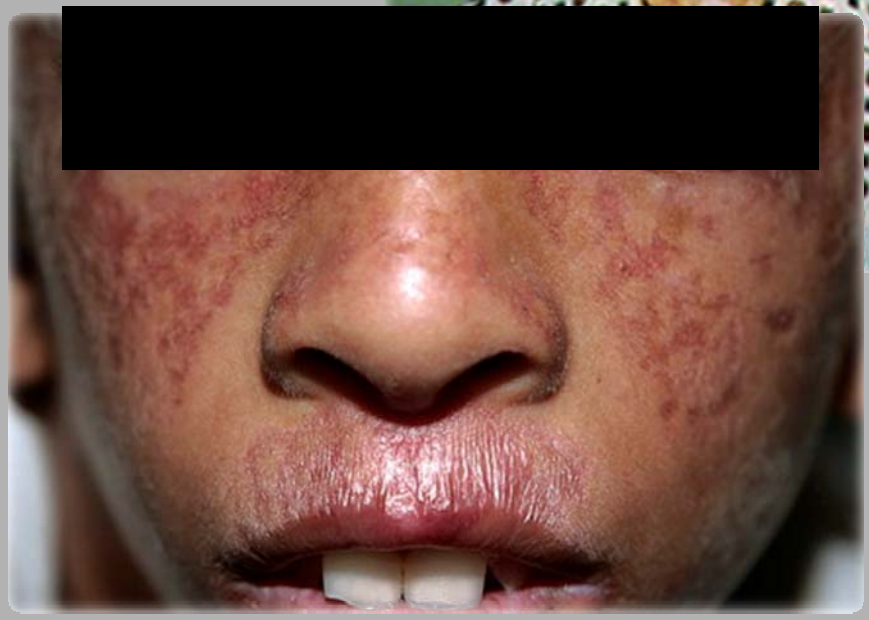
Bloom syndroom

Huidafwijkingen







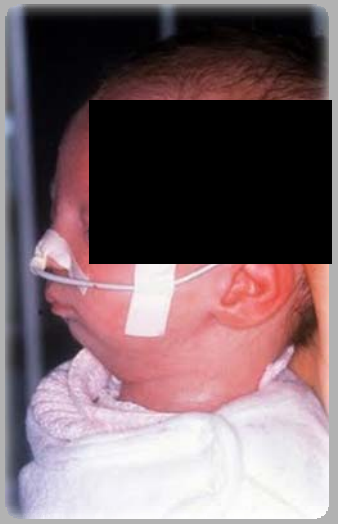


Bloom syndroom

Systemische afwijkingen

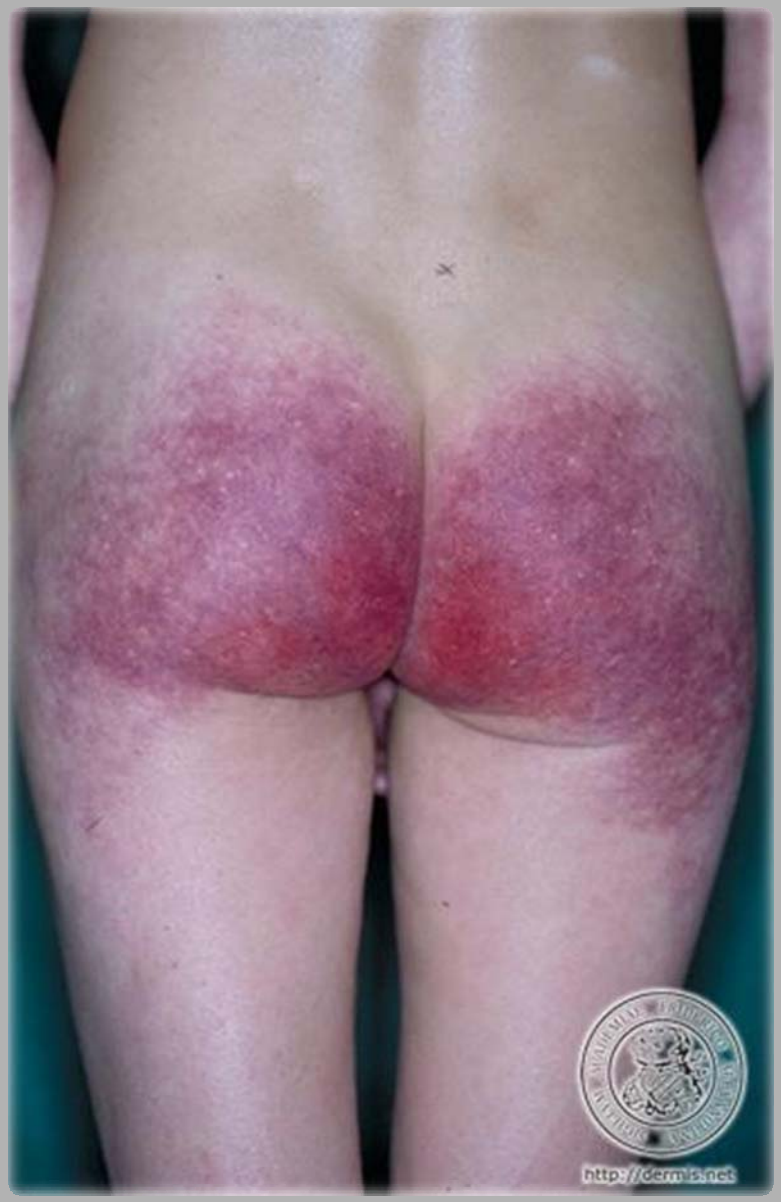
Maligniteit:

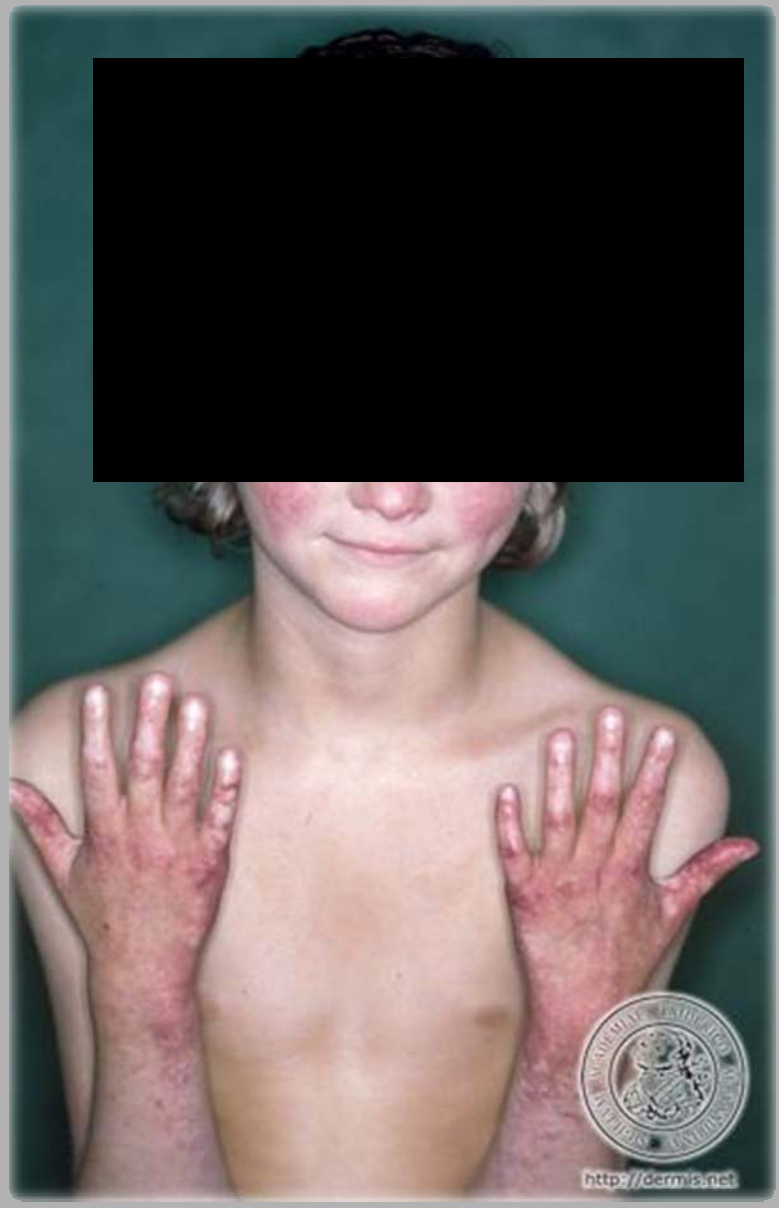
- Leukemie
- Lymfomen
- Neuroblastomen
- GI-tumoren



Rothmund-Thomson syndroom

Huidafwijkingen


http://dermis.net

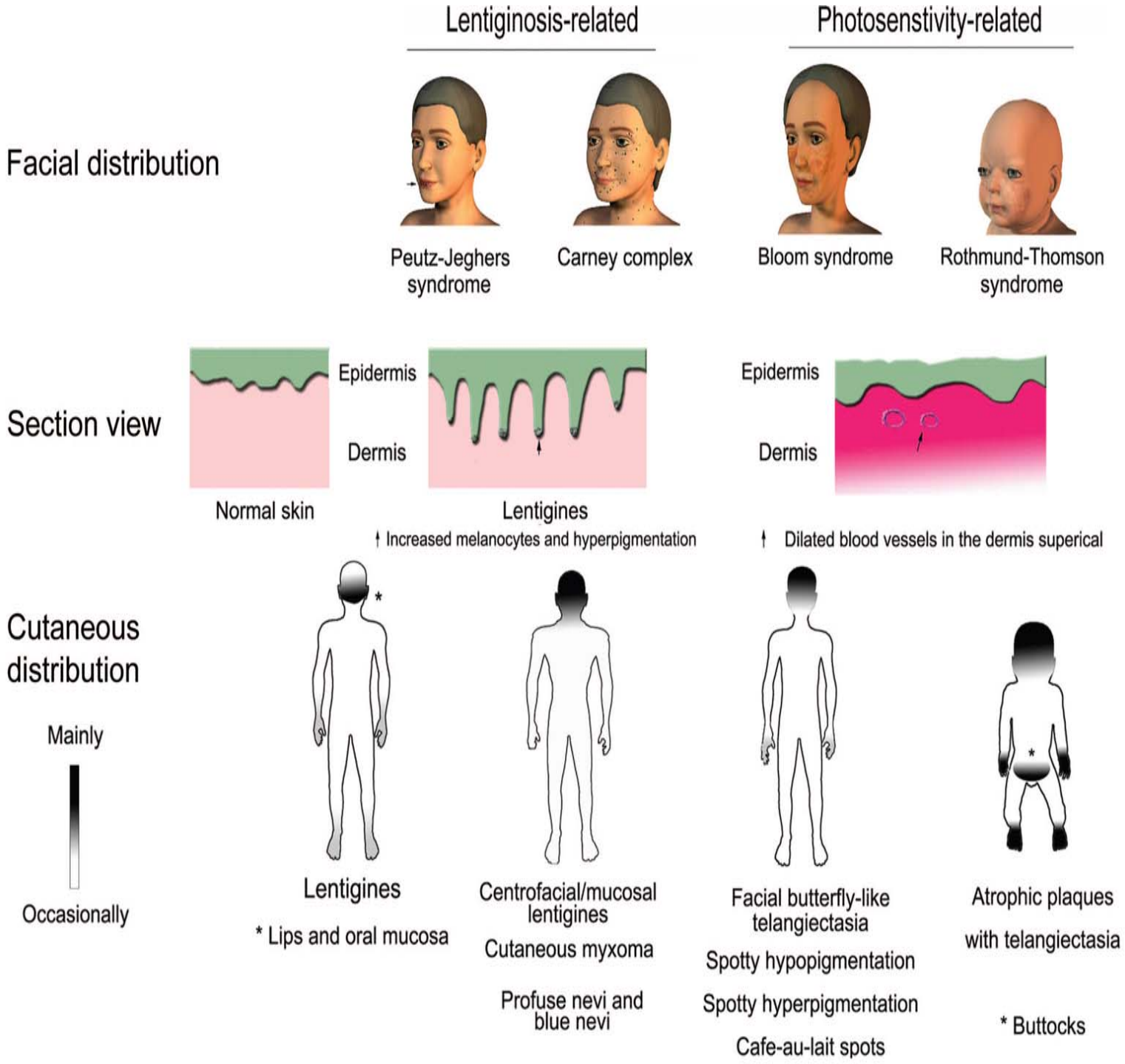

http://dermis.net

Rothmund-Thomson syndroom

Systemische afwijkingen

Maligniteit

- Osteosarcoom
- Basaalcelcarcinoom huid


Lentiginosis-related
Photosenstivity-related
Facial distribution
Peutz-Jeghers syndrome
Carney complex
Bloom syndrome
Rothmund-Thomson syndrome
Section view
Epidermis
Dermis
Normal skin
Lentigines
Increased melanocytes and hyperpigmentation
Epidermis
Dermis
Dilated blood vessels in the dermis superical
Cutaneous distribution
Mainly
Occasionally
*
Lentigines
* Lips and oral mucosa
Centrofacial/mucosal lentigines
Cutaneous myxoma
Profuse nevi and blue nevi
Facial butterfly-like telangiectasia
Spotty hypopigmentation
Spotty hyperpigmentation
Cafe-au-lait spots
*
Atrophic plaques with telangiectasia
* Buttocks

Hamartoom gerelateerde aandoeningen

- Genetische afwijking
- Tumorgroei

Voorbeelden:

- Cowden syndroom
- Multiple Endocriene Neoplasma syndroom

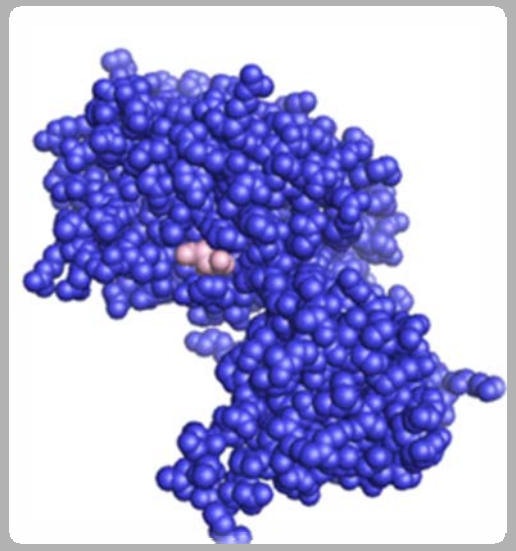
MEN-1/ MEN-2B

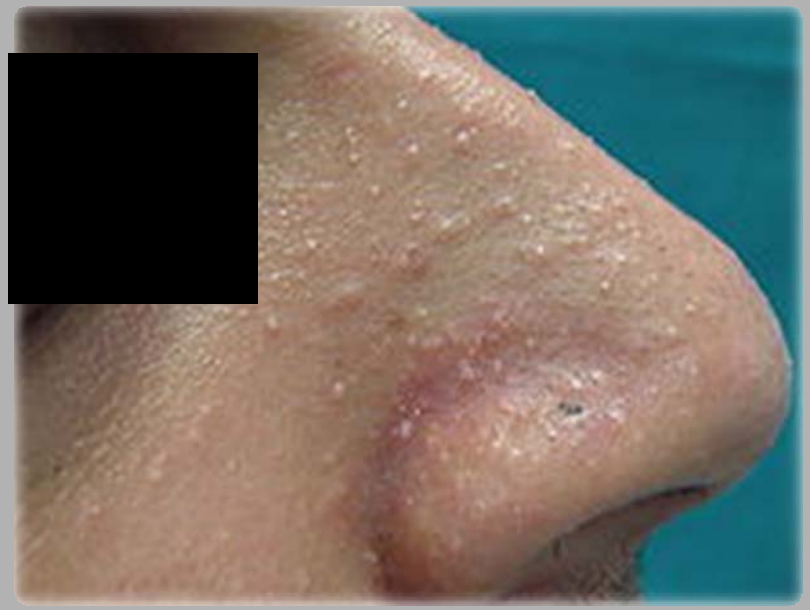
- Tuberous Sclerosis Complex (TSC)
- Gardner syndroom
- Muir-Torresyndroom (MTS)

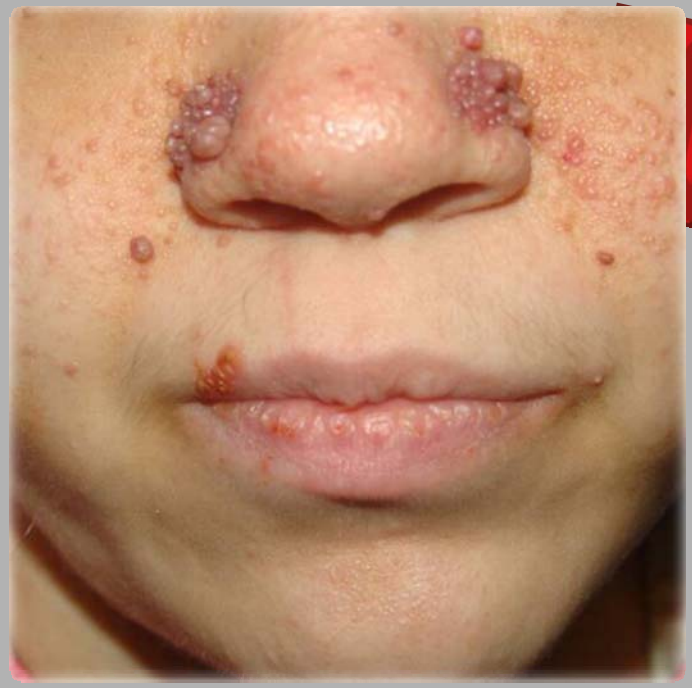
Cowden syndroom

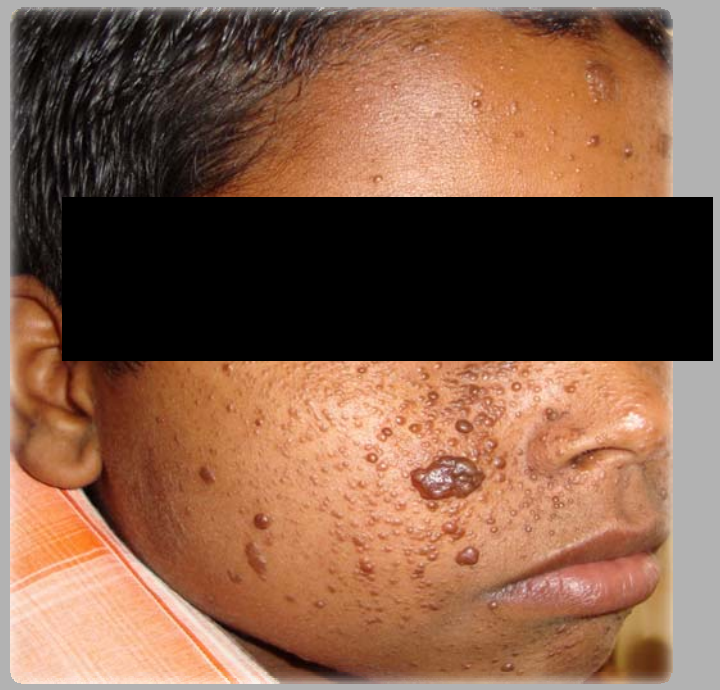
PTEN- chromosoom afwijking

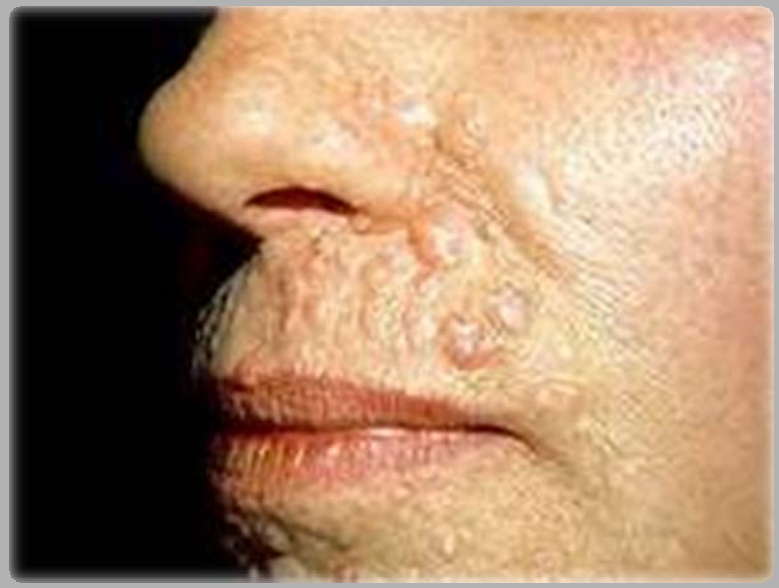
Huidafwijking











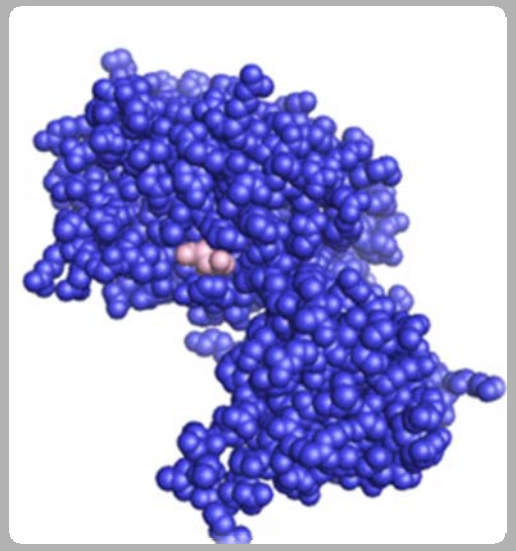
Cowden syndroom

Systemische afwijkingen

Maligniteit

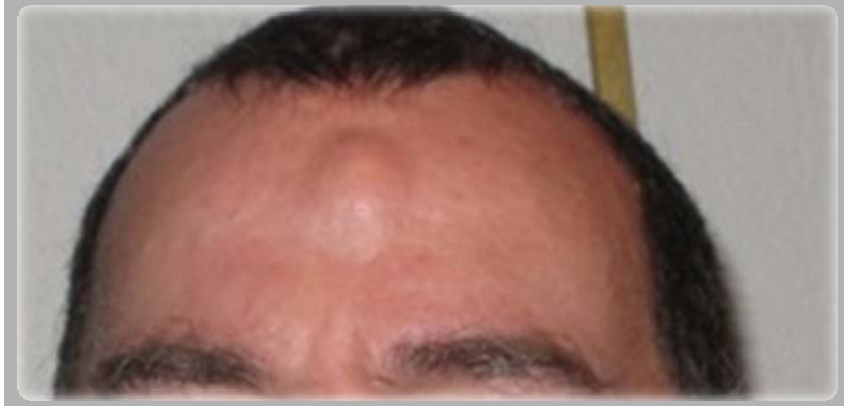
- Mamma
- Schildklier
- Endometrium

Richtlijn oncoline

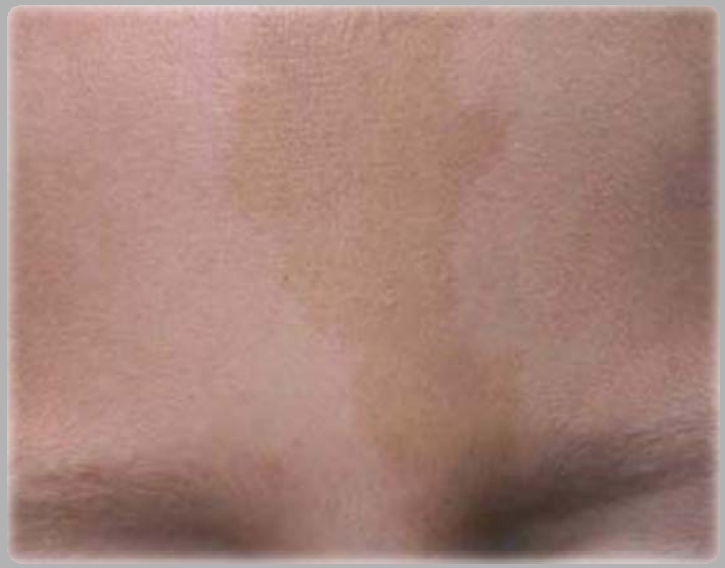


MEN-1/ 2B syndroom

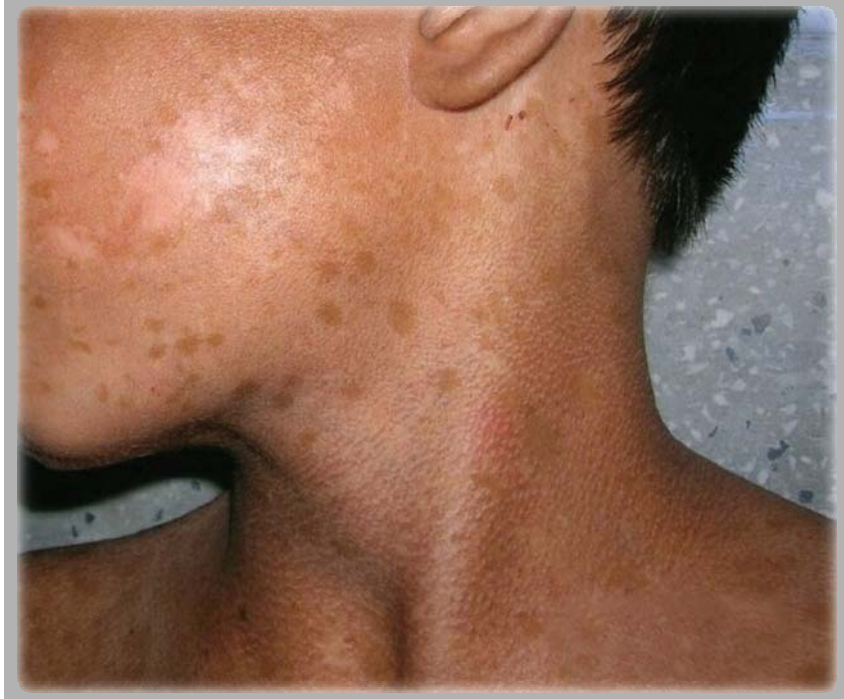
Huidafwijkingen (verschil MEN 1 of MEN 2b)



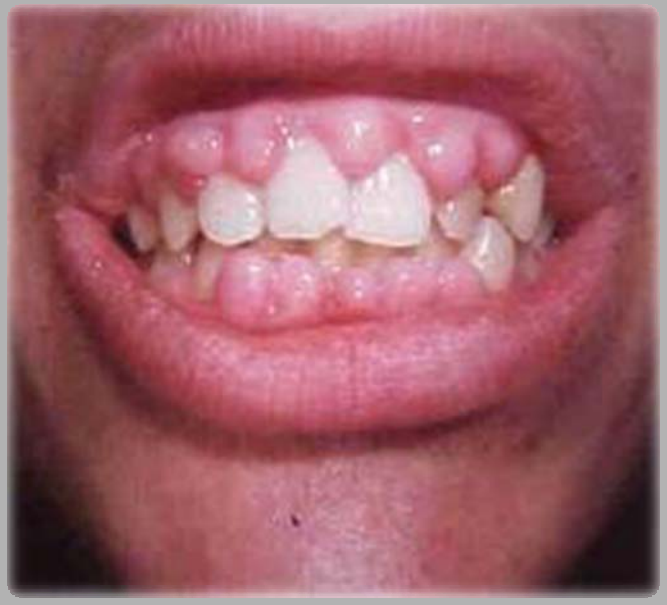
Subcutaan lipoom



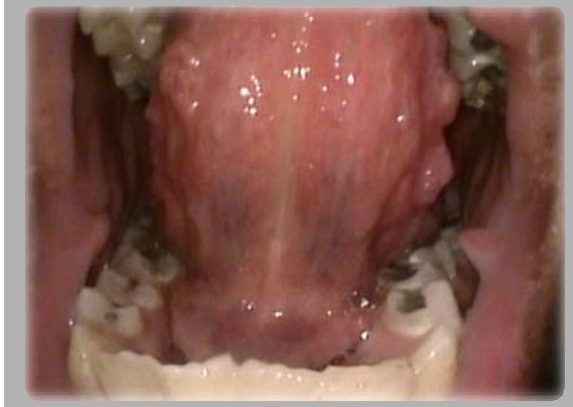
Café au lait macule

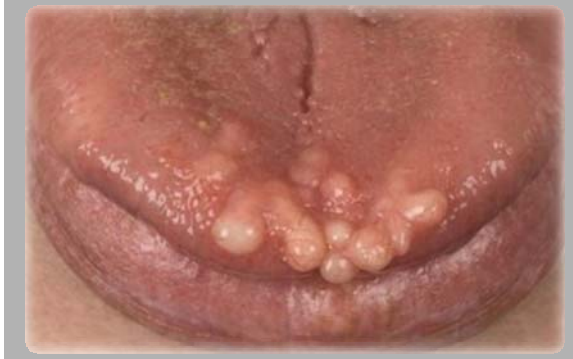


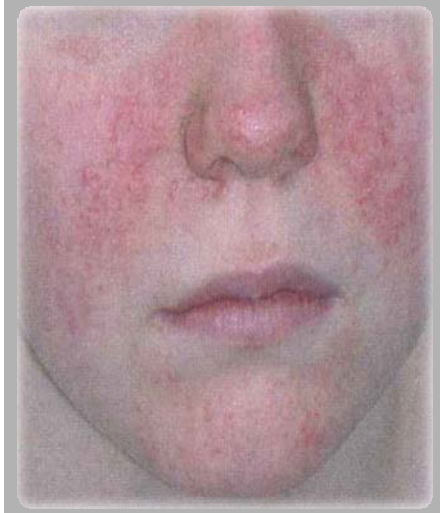
Confetti-achtige maculae

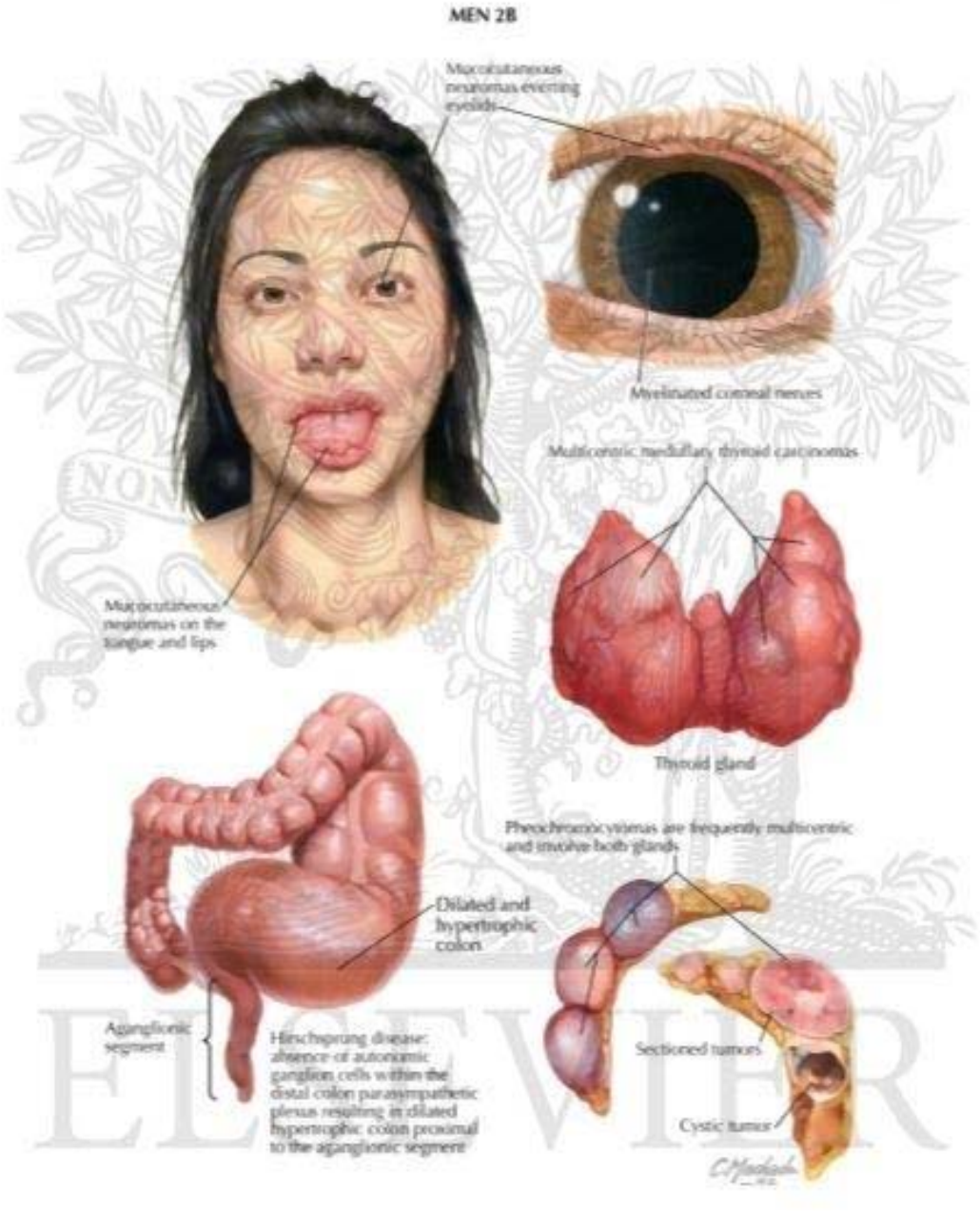


Tandvleespoliepen









MEN-1/ 2B syndroom

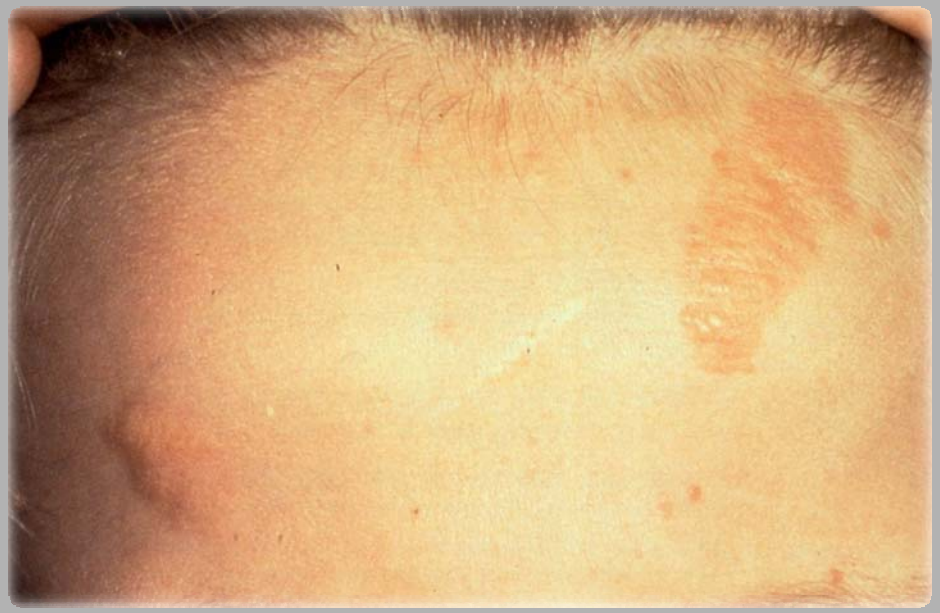
Systemische afwijkingen

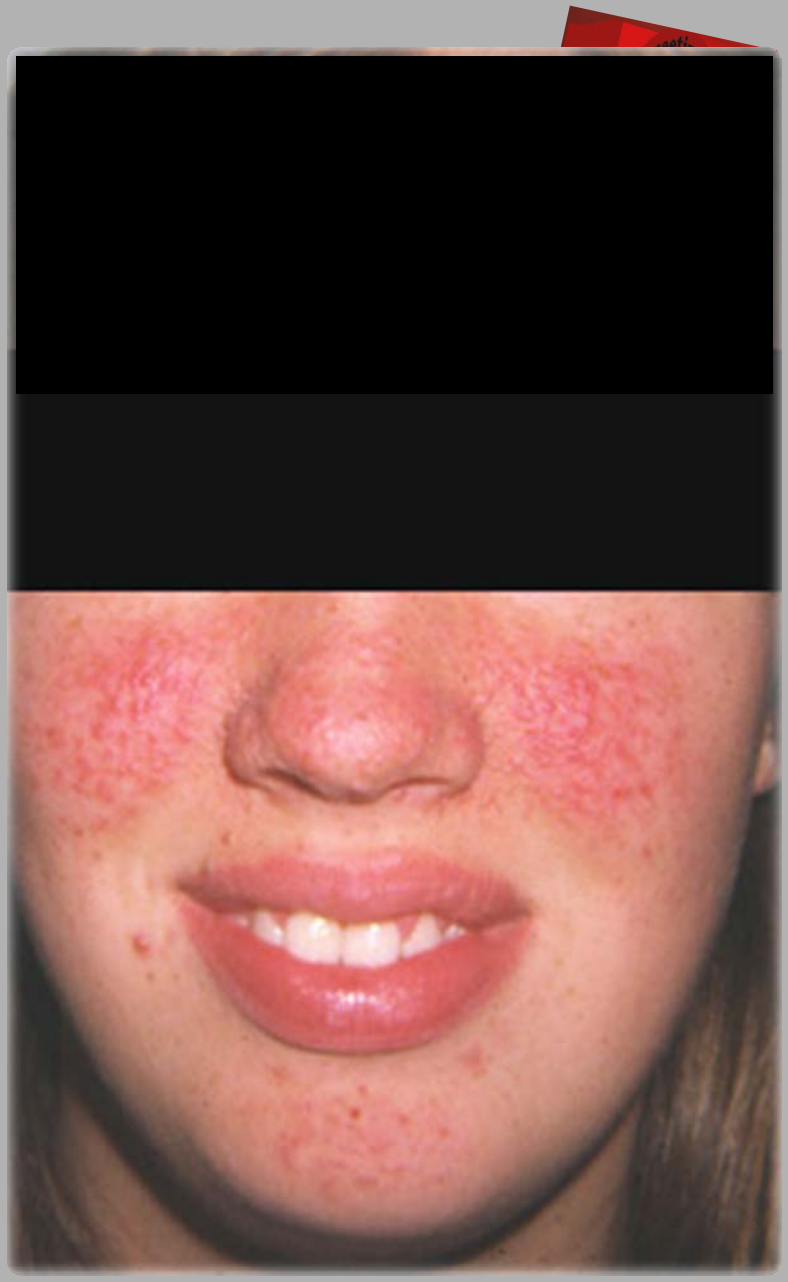
Maligniteit:

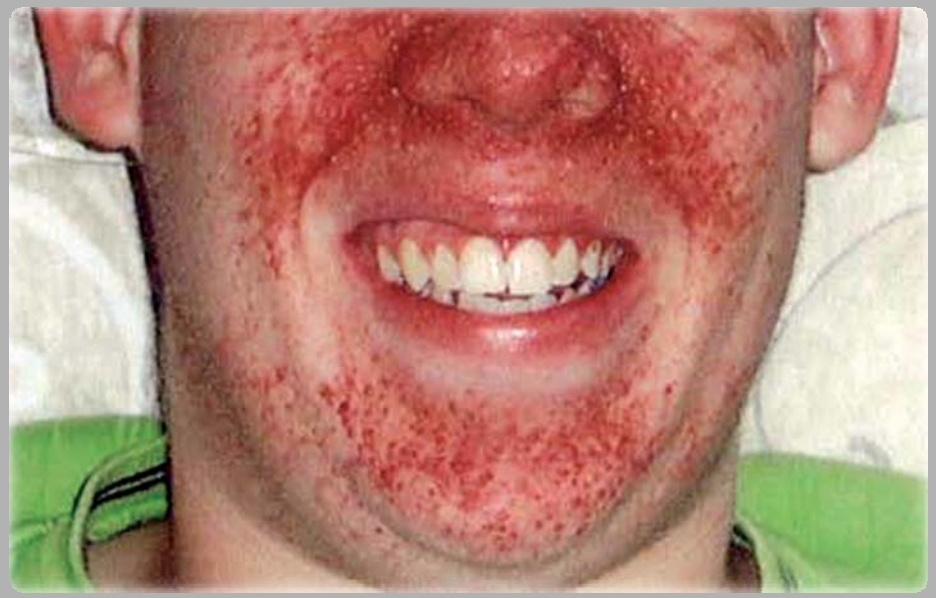
- Bestaande tumorgroei ontaart maligne

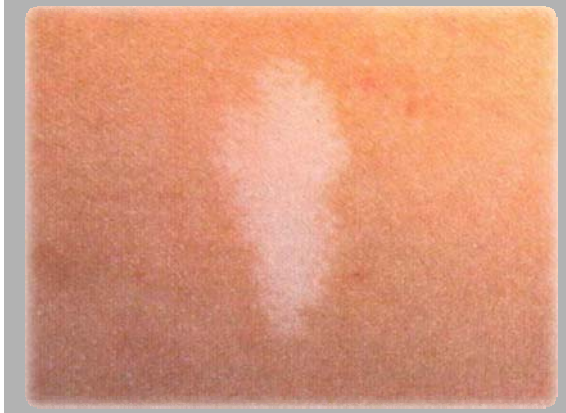
Tuberous Sclerosis Complex (TSC)

Huidafwijkingen

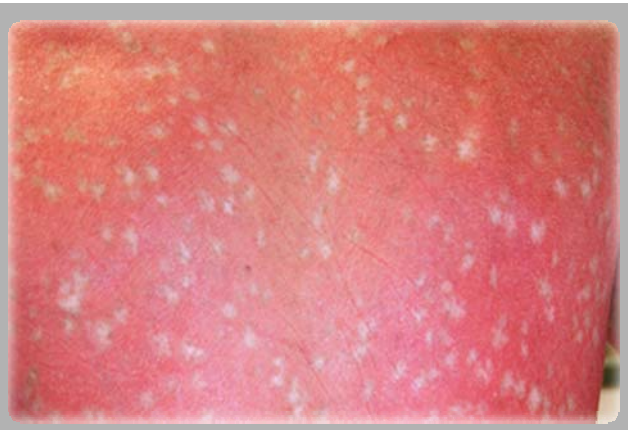




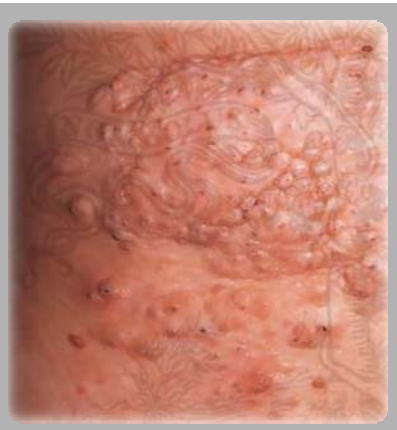




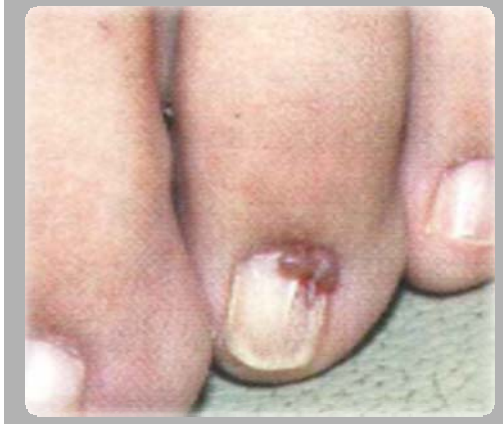
Hypomelanotic macule (ash leaf spot)



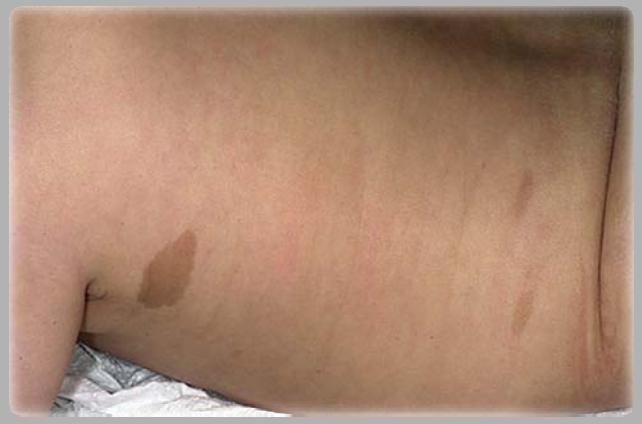
Confetti-like spots



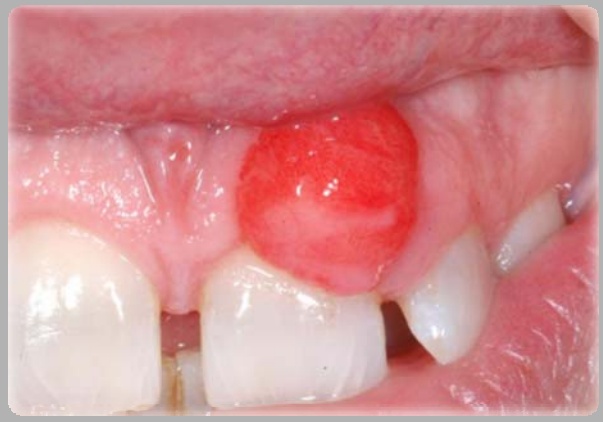
Shagreen patch



Periungual fibromas



Café au lait spots



Gingival fibroma

Tuberous Sclerosis Complex (TSC)

Systemische afwijkingen

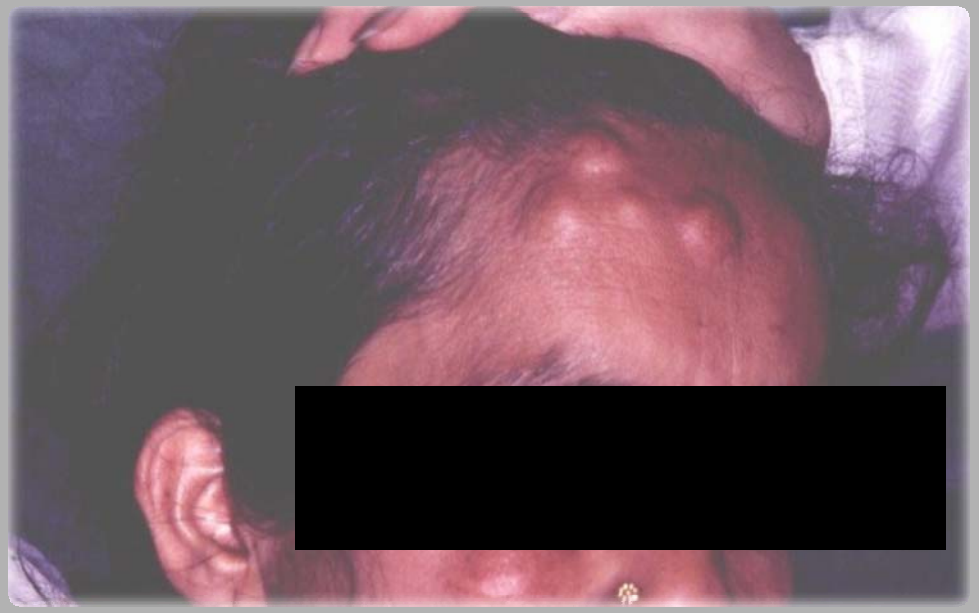
Maligniteit:

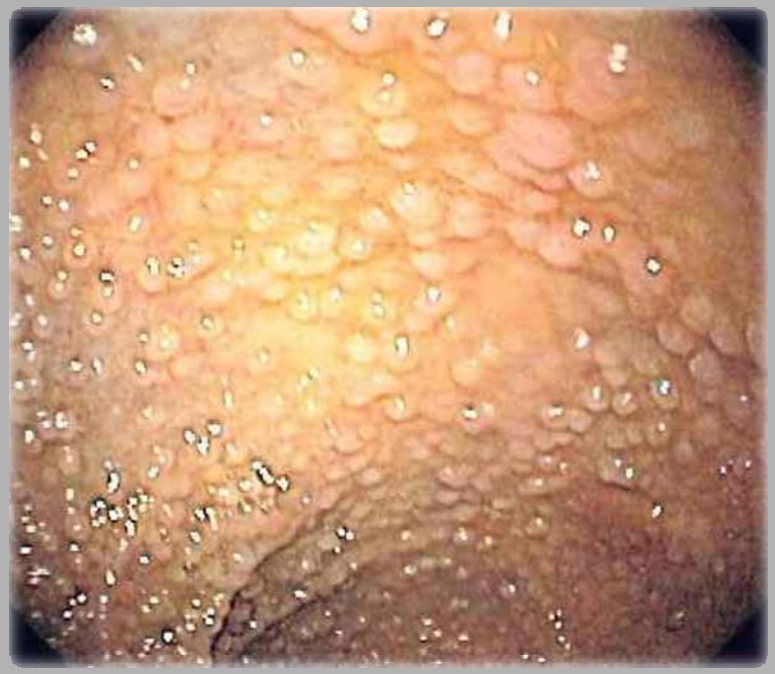
- Rhabdomyoom
- Niercelcarcinoom
- Hersentumoren

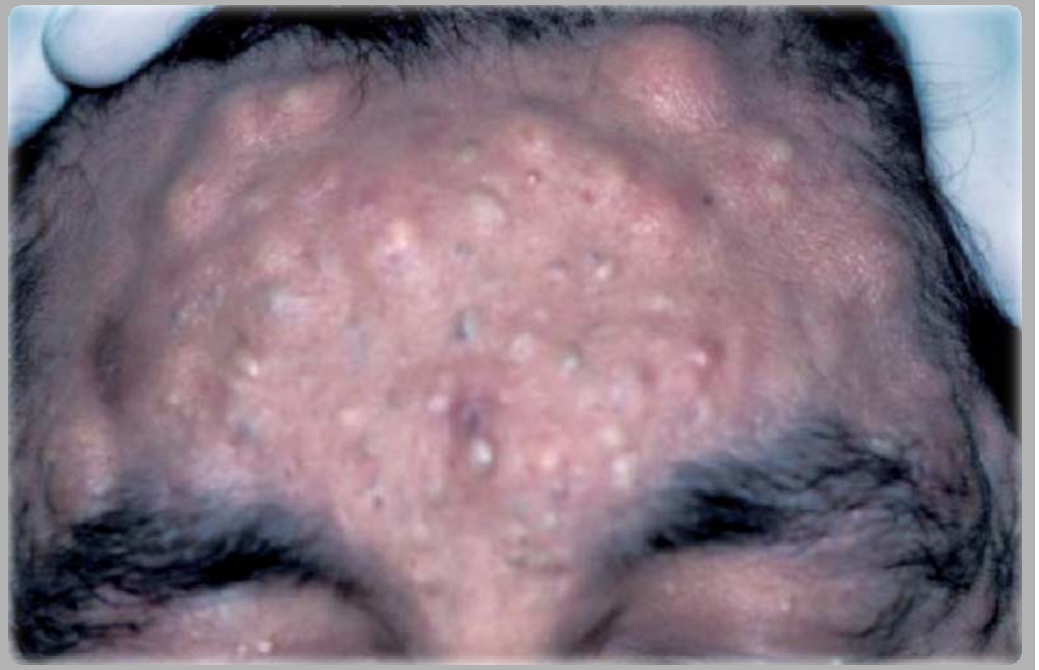
Gardner syndroom

Mutatie van het APC-gen

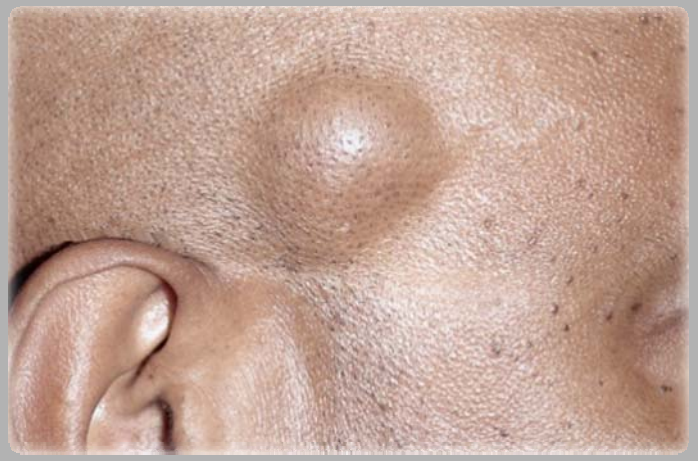
Huidafwijkingen



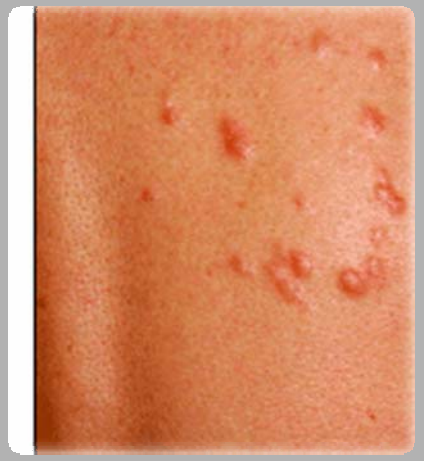




Fibroma

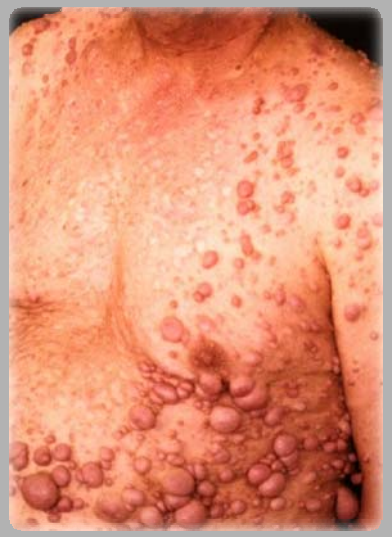


Lipoma

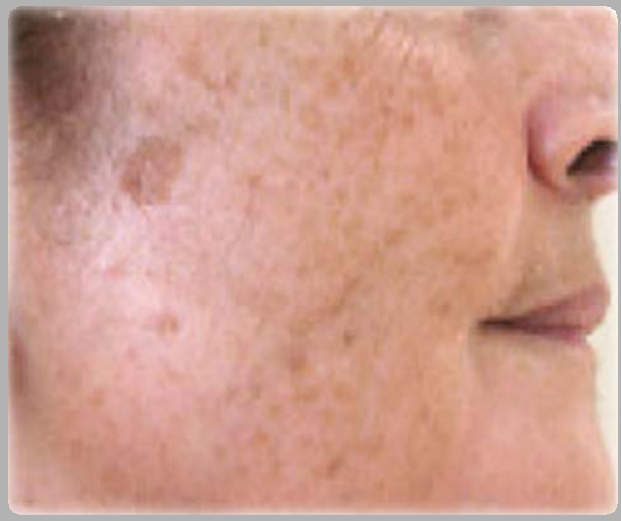


Leiomyoma

Pilomatricomas



Neurofibromas



Pigmented skin lesions

Gardner syndroom

Systemische afwijkingen

Maligniteit:

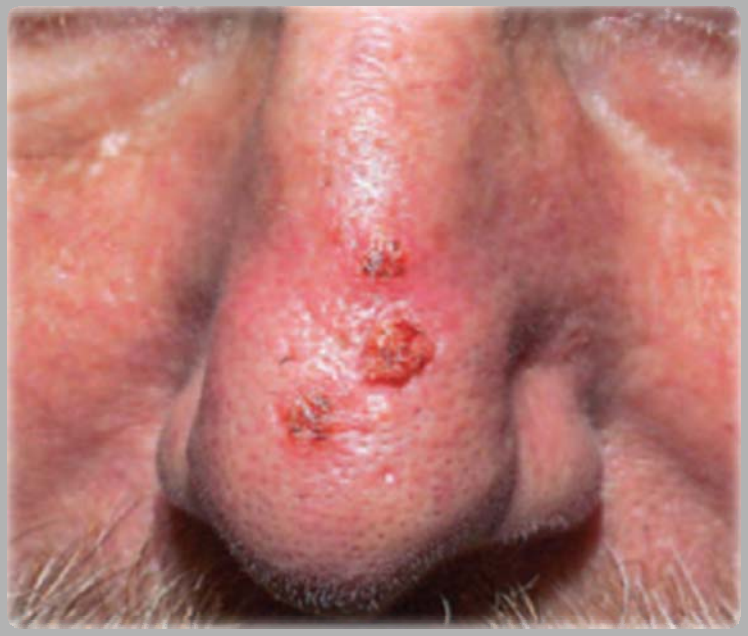
- Coloncarcinoom
- Schildkliertumoren
- Desmoïdtumoren

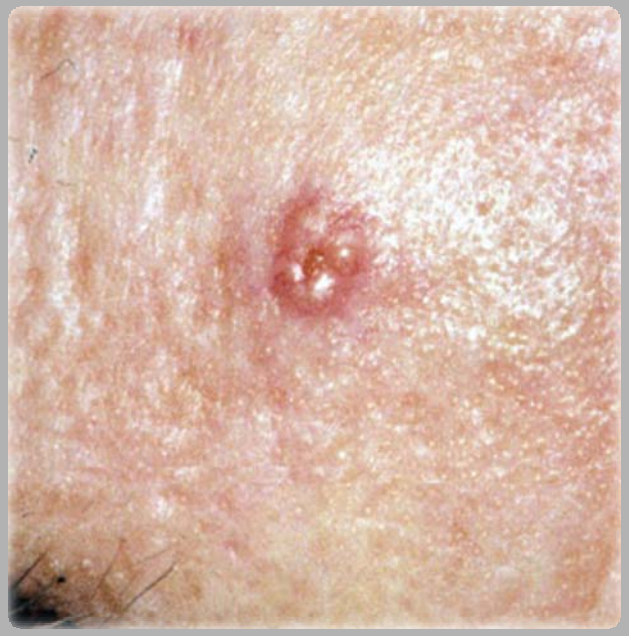
Muir-Torresyndroom (MTS)

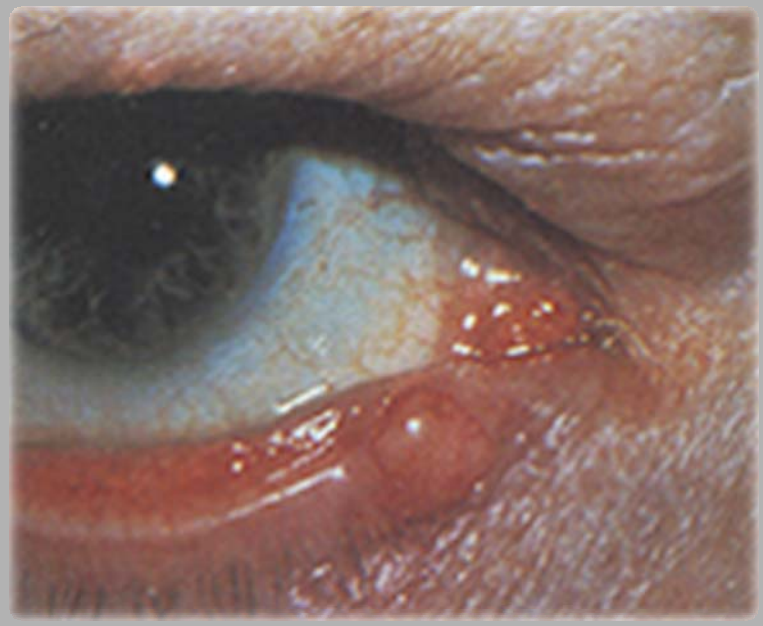
Afwijking in het MSH2 gen

Presentatie op volwassen leeftijd

Huidafwijkingen







Muir-Torresyndroom (MTS)

Systemische afwijkingen

Maligniteit:

- Coloncarcinoom
- Urogenitale maligniteit
- Hematologische maligniteit
- Uterus/ Ovarium
- Hoofd-hals

Hamartoma-related

Facial distribution


Cowden syndrome



Multiple endocrine neoplasia syndrome 1 and 2B

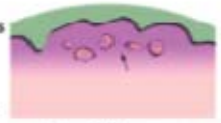

Tuberous sclerosis complex


Gardner syndrome


Muir-Torre syndrome

Section view

Trichilemmoma
- Derived from the outer root sheath epithelium of hair follicle



Angiofibroma
- Fibrous and vascular proliferation

Neuroma
- Encapsulated mass of nerve cells and tissues

Angiofibroma
- Fibrous and vascular proliferation

Epidermoid cyst
- Anucleate keratinaceous contents lined by thin wall of squamous epitheliums

Sebaceous neoplasms
- Nodular lobulated growth with sebaceous differentation

Cutaneous distribution

Mainly

Occasionally


Facial follicular hamartomas

Multiple skin tags

Acral keratoses

Palmoplantar keratoses


Multiple facial angiofibromas

Subcutaneous lipomas

Collagenomas

Café-au-lait macules


Neuromas of lips and tongue

Thick lips


Multiple facial angiofibroma

White leaf-shaped macules

Ungual or subungual fibromas

Shagreen patches

Café-au-lait spots


Epidermoid cysts

Desmoid tumors

Fibromas

Lipomas

Leiomyomas

Pilomatricomas


Sebaceous gland neoplasms

Keratoacanthomas

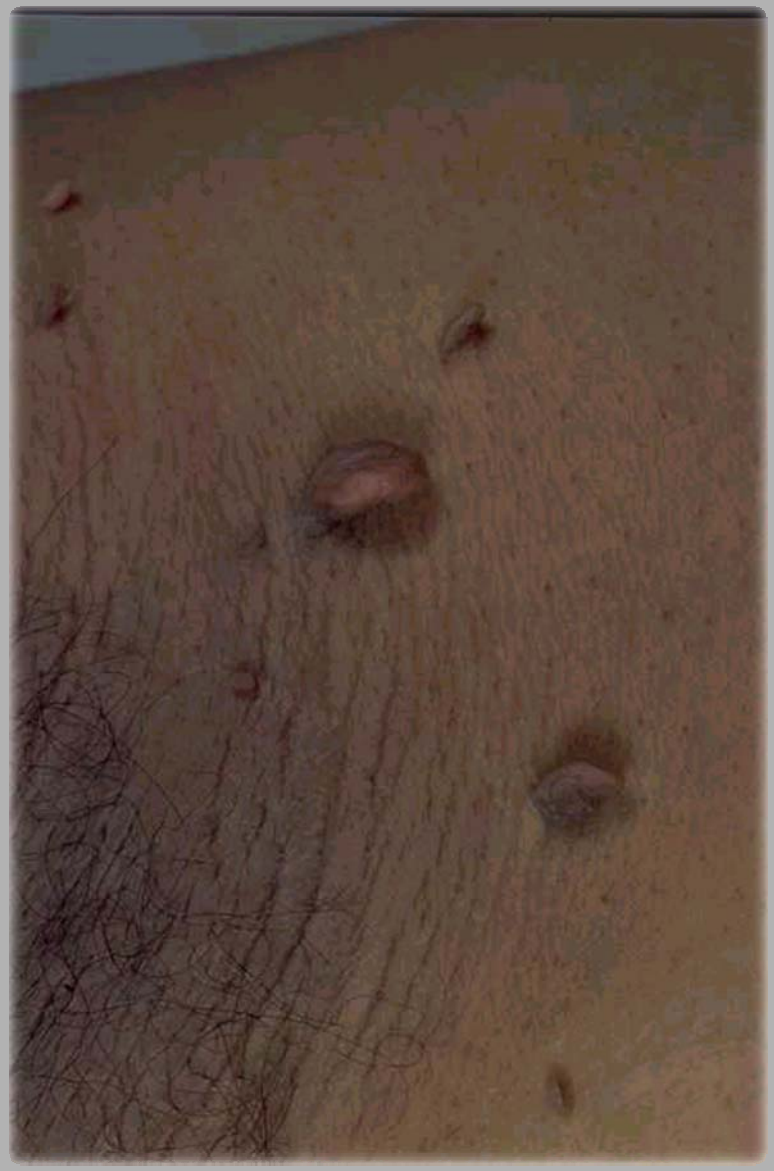
Basal cell carcinoma

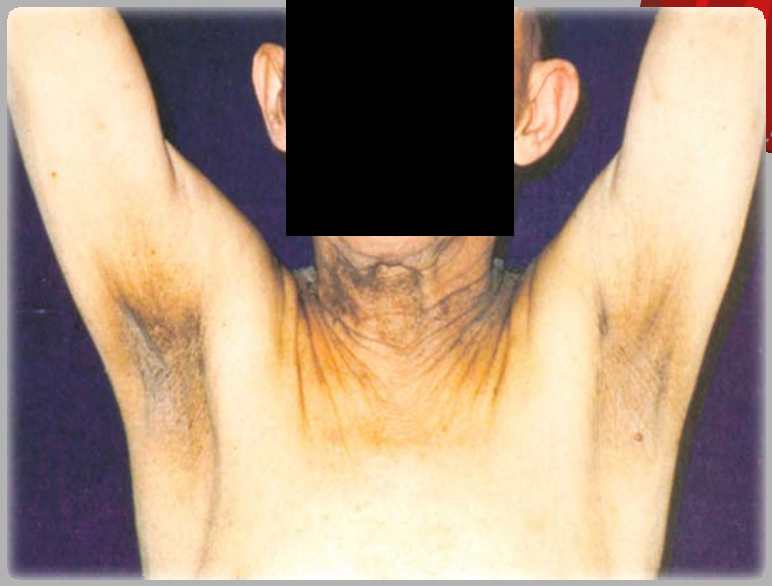
Overige aandoeningen

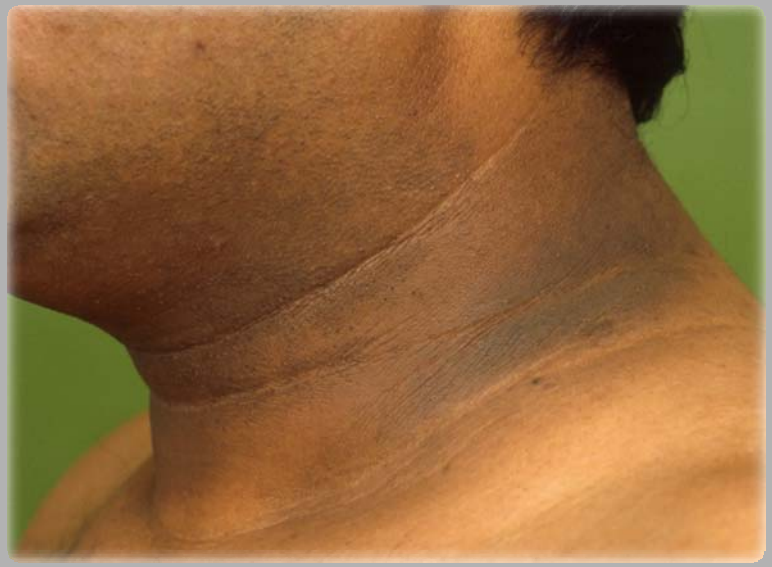
- Paraneoplastische syndromen
- Niet erfelijk bepaald
- Vaak geen systemische afwijkingen
- Sterke associatie met maligniteit

Acanthosis Nigricans

Huidafwijking







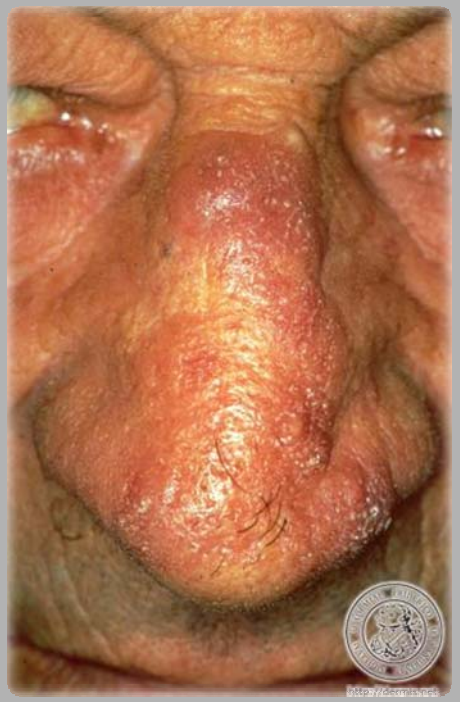
Acanthosis Nigricans

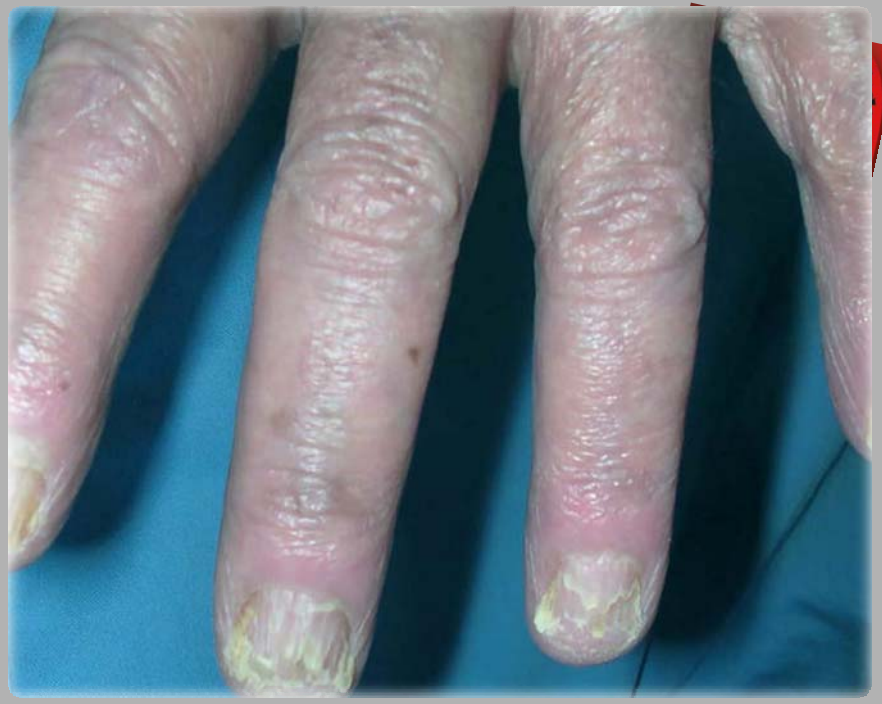
Maligniteit:

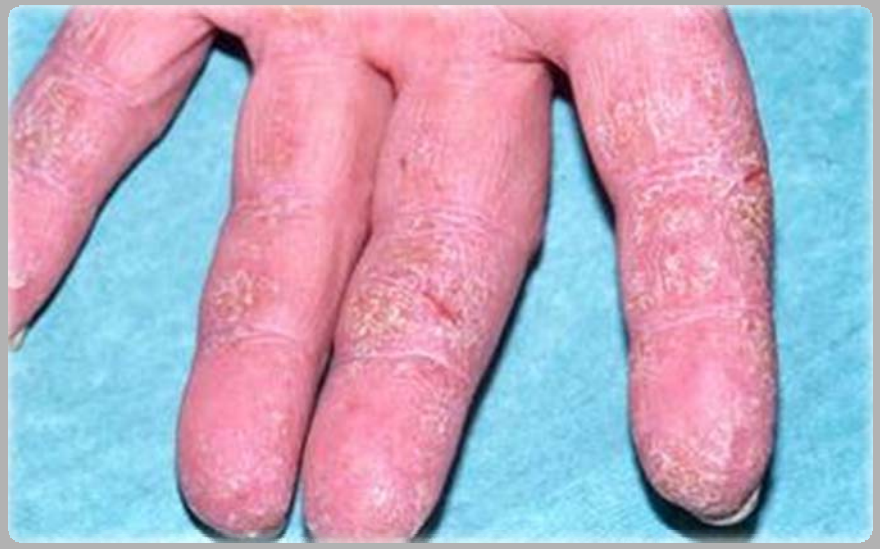
- GI-traject

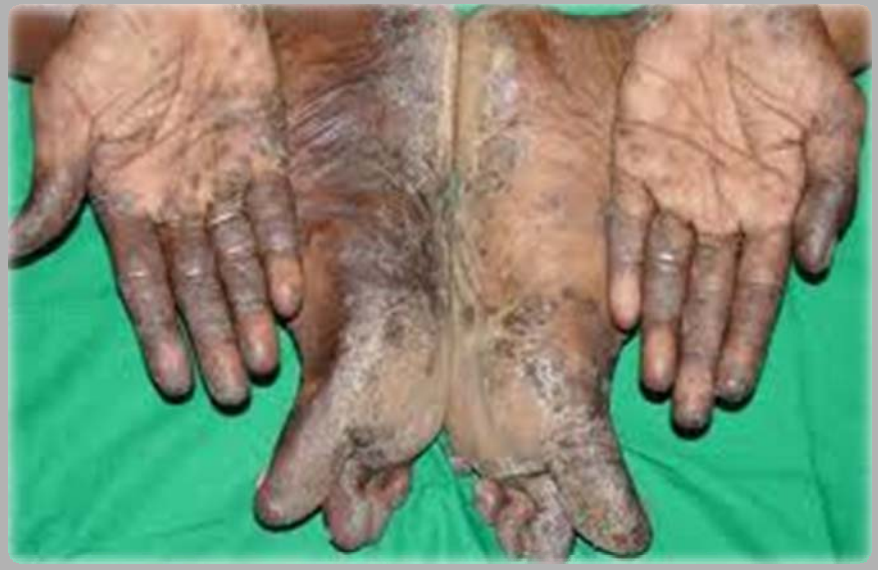
Paraneoplastische acrokeratosis van Bazex

Huidafwijkingen









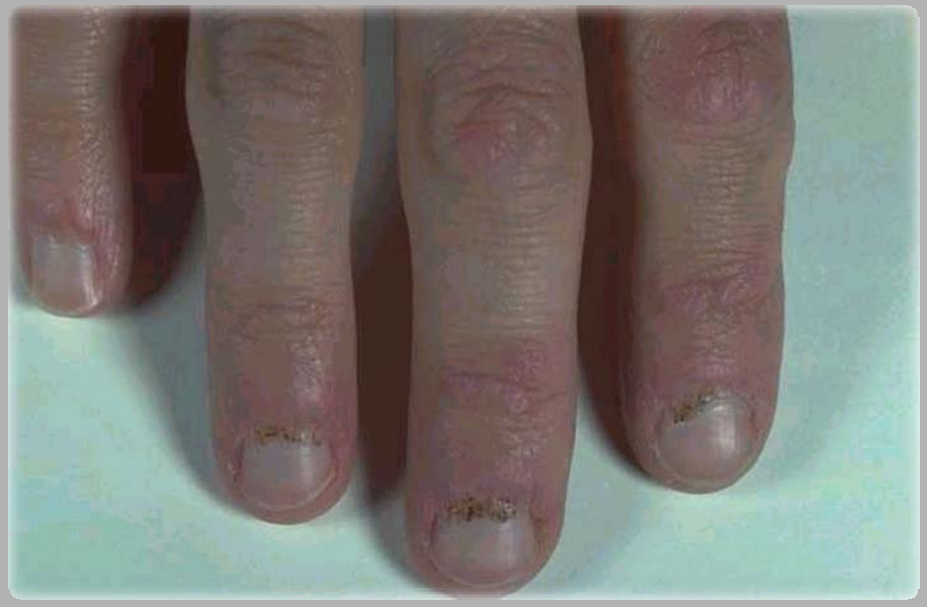
Paraneoplastische acrokeratosis van Bazex

Maligniteit

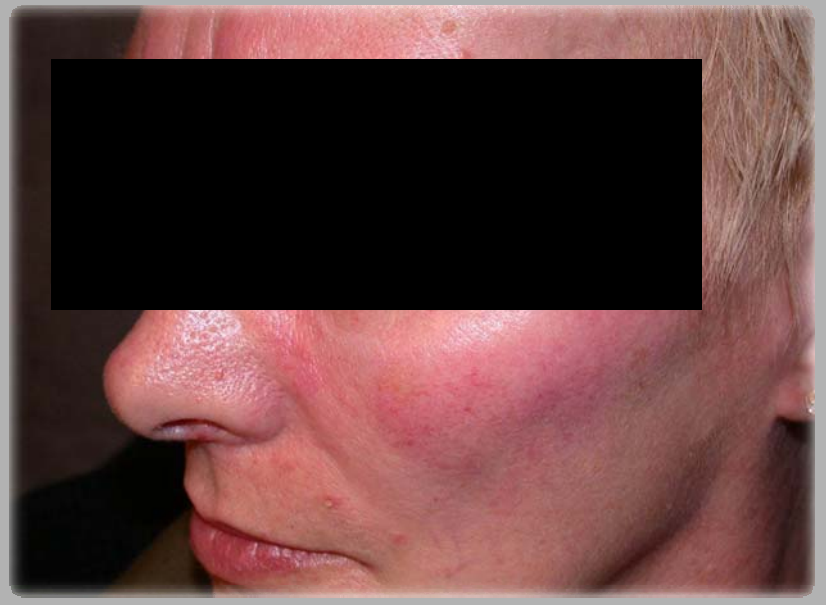
- Bijna 100%
- Hoofd-hals
- Long
- Darmen

Dermatomyositis

Huidafwijking



Gottronse papels



Heliotrope rash

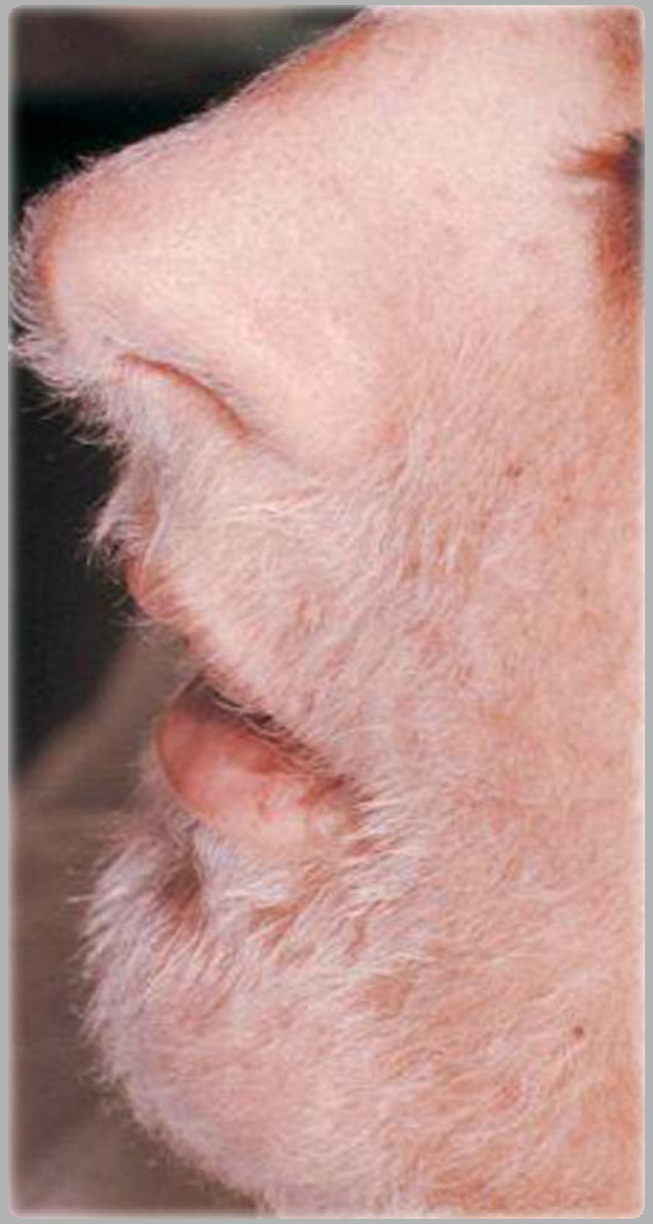
Dermatomyositis

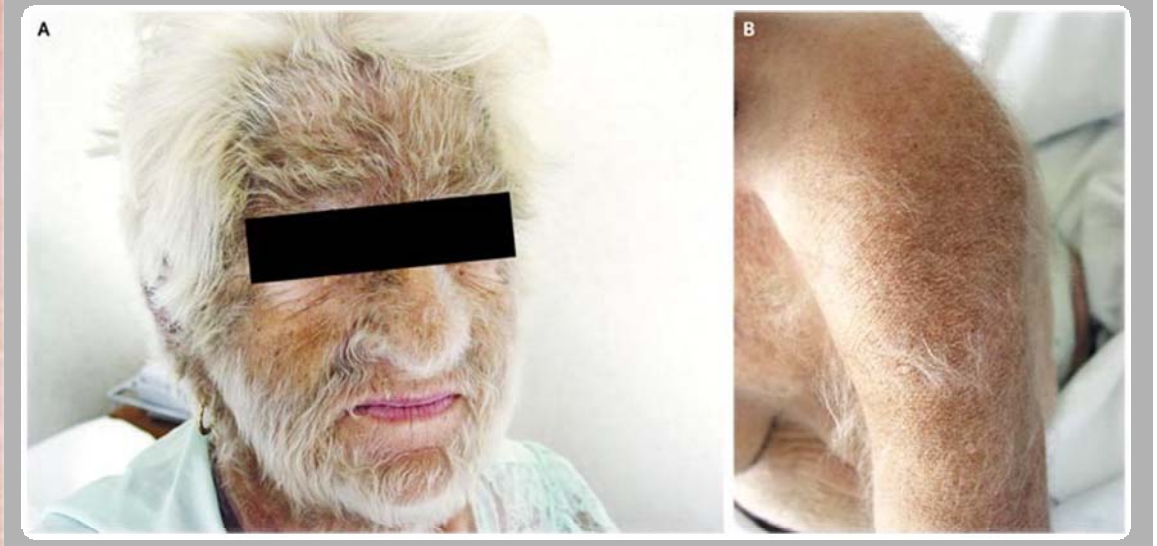
Maligniteit:

- In 25% associatie met een maligniteit
- Ovarium
- Cervix
- GI-traject

Hypertrichosis lanuginosa acquisita (HLA)

Huidafwijking




A
B

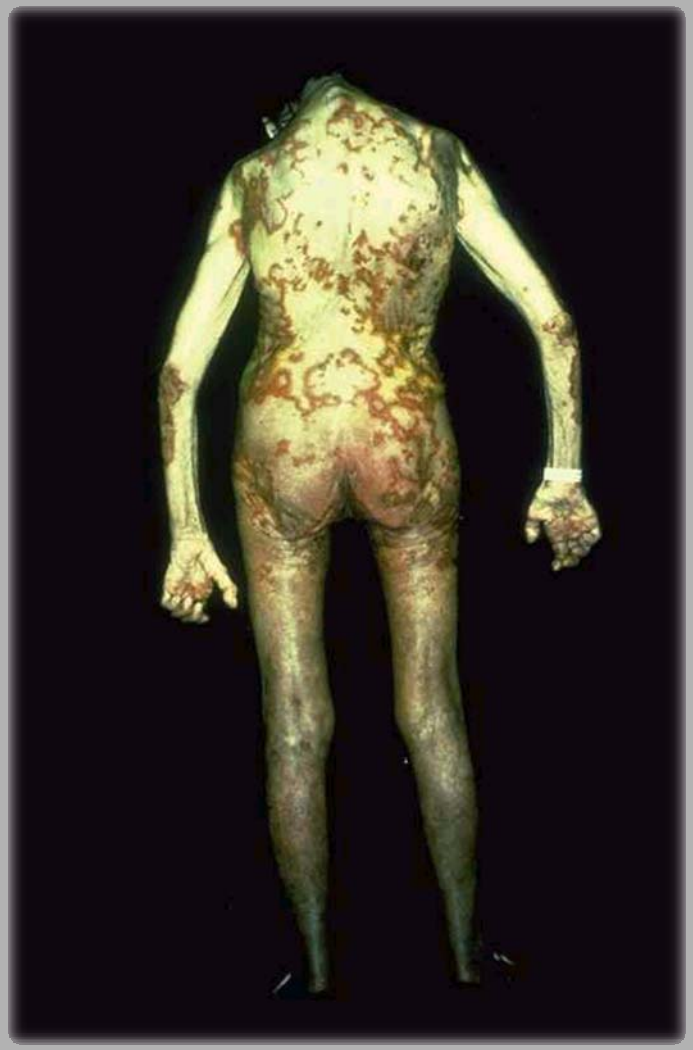
Hypertrichosis lanuginosa acquisita (HLA)

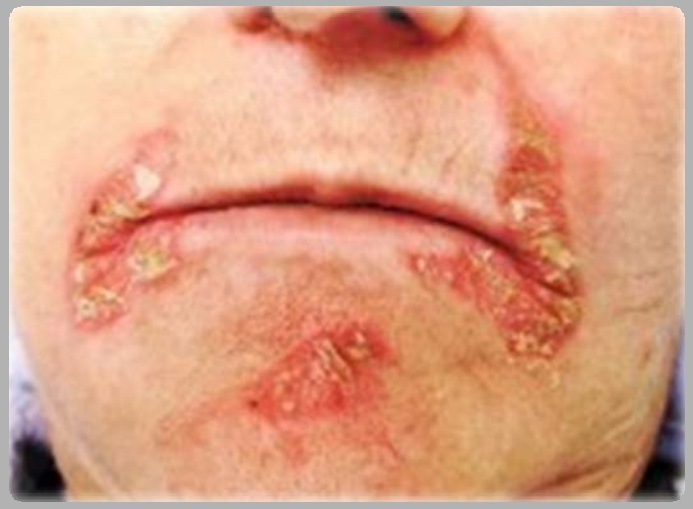
Maligniteit:

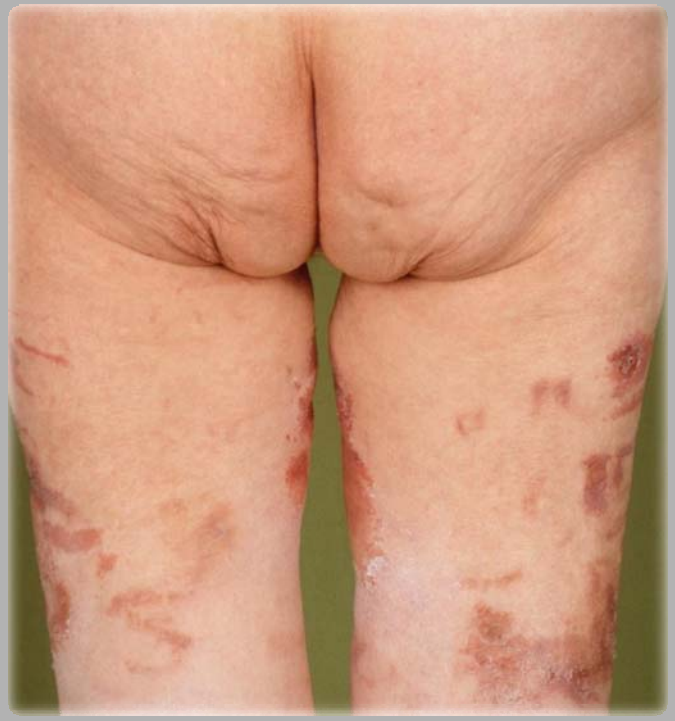
- Bij >75%
- Long
- Colon
- Uterus
- Lymfoom

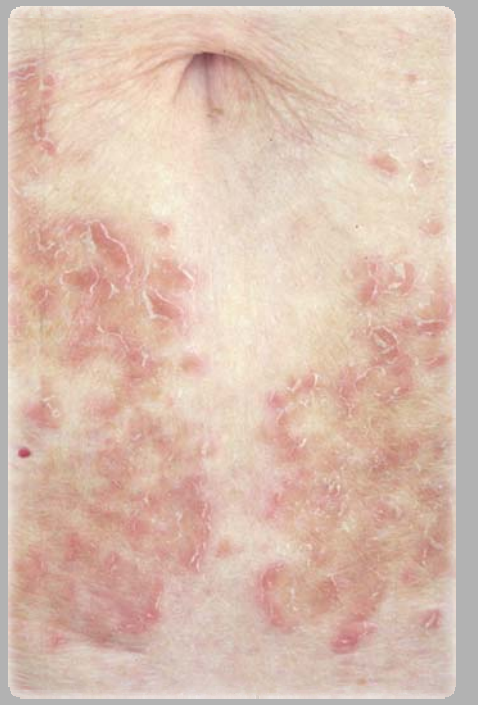
Necrolytisch migrerend erytheem

Huidafwijking









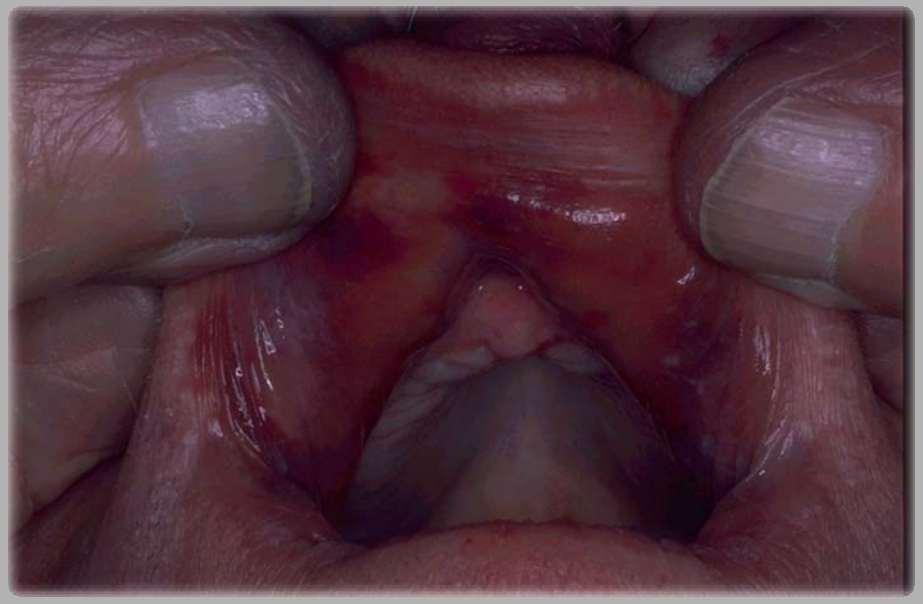
Necrolytisch migrerend erytheem

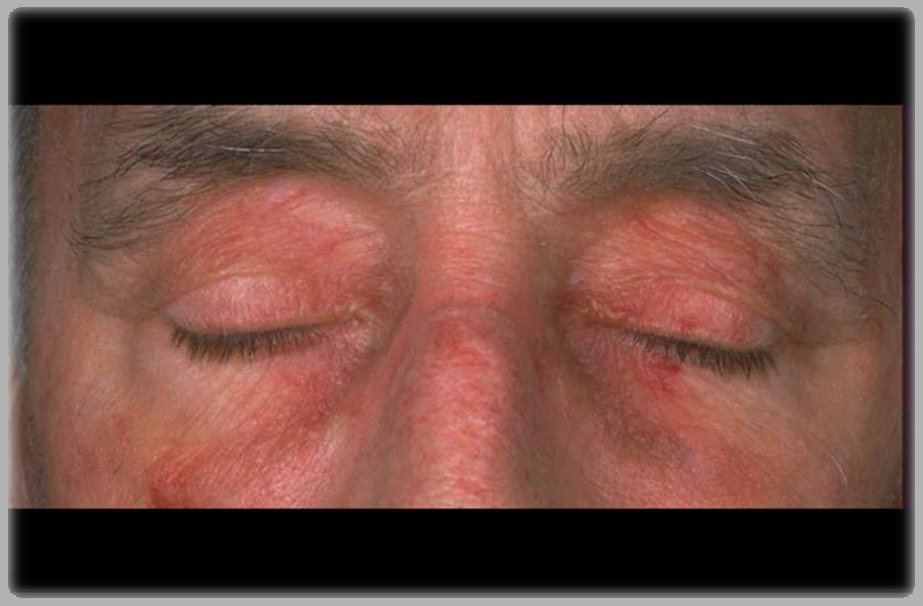
Maligniteit:

- Pancreas
- Lever
- Longen
- en/of duodenum

Amyloidose

Huidafwijking





Cutane amyloidose

Amyloidose

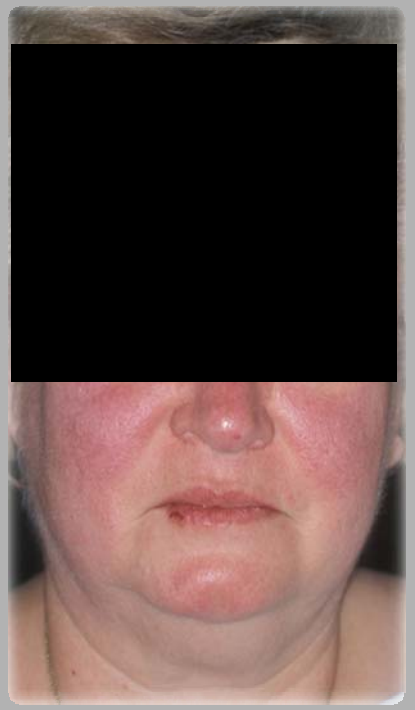
Maligniteit:

- Multipel Myeloom

Carcinoid syndroom

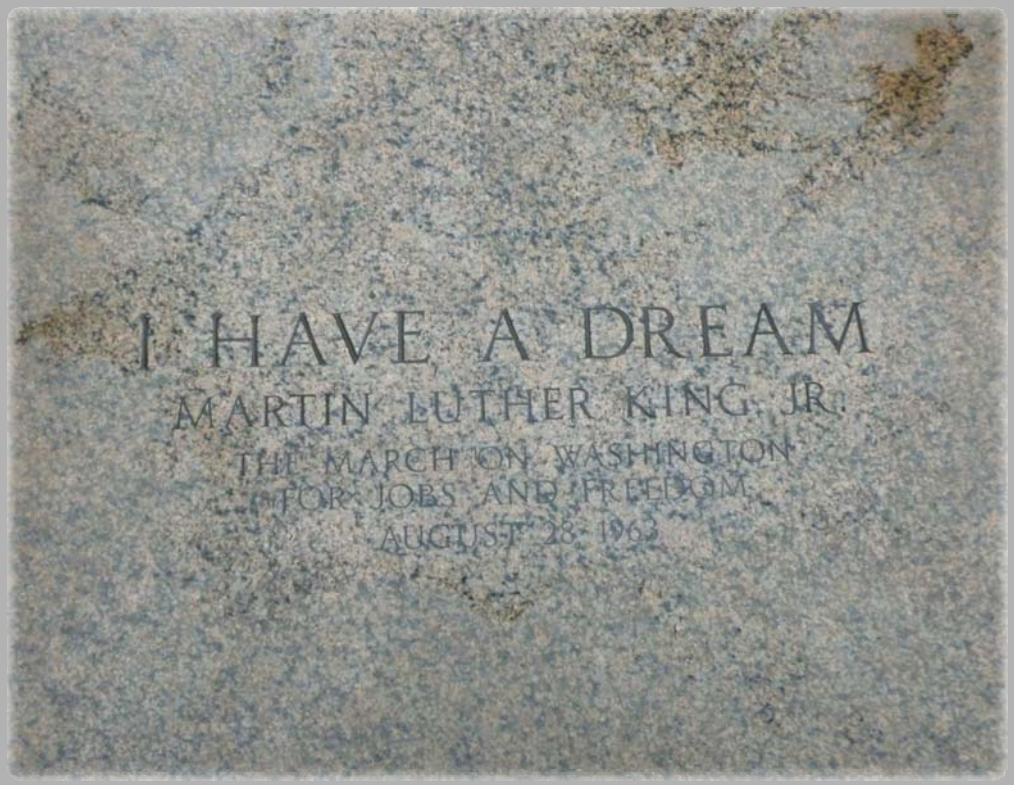
- Blozen, meestal 10-30 minuten
- Vaak in combinatie met andere klachten
- Bij 75%

- Oorzaak: hormonaal



Vragen, opmerkingen, etc.

nschrama@elkerliek.nl



Handige websites (1)

- Genetic Alliance:

http://www.geneticalliance.org

- National Organization for Rare Disorders (NORD)

http://www.rarediseases.org/

- Peutz Jegher USA

http://ghr.nlm.nih.gov/condition/peutz-jeghers-syndrome

- Peutz Jegher Nederland

http://www.oncoline.nl/peutz-jeghers-syndroom

- Huidziekten

www.huidziekten.nl

- Carney Complex

http://ghr.nlm.nih.gov/condition/carney-complex

- Bloom syndroom

http://www.bloomsyndrome.eu/dutch/whatis.php

Handige websites (2)

- Rothmund-Thomson

http://www.medisuv.com/nl/1112.html

- Cowden syndroom

http://pten.nl/syndroomcowden/

- MEN-1 syndroom

http://www.men.nfk.nl/ziektebeschrijvingen/men1

http://www.men.nfk.nl/ziektebeschrijvingen/men2b

- Tuberous sclerosis complex (tsc)

https://nl.wikipedia.org/wiki/Tubereuze_sclerose_complex

- Muir-Torre Syndroom

http://www.cancer.net/cancer-types/muir-torre-syndrome

- Carney Complex

http://ghr.nlm.nih.gov/condition/carney-complex

Literatuurlijst (1)

- Bartsch, D.K., Gress, T.M., & Langer, P. (2012). Familial pancreatic cancer□□current knowledge. Nature Reviews. Gastroenterology & Hepatology, 9, 445□453.
- Baskin, H.J. (2008). The pathogenesis and imaging of the tuberous sclerosis complex. Pediatric Radiology, 38, 936□952.
- Borkowska, J., Schwartz, R.A., Kotulska, K., & Jozwiak, S. (2011). Tuberous sclerosis complex: Tumors and tumorogenesis. International Journal of Dermatology, 50, 13□20.
- Capurso, G., Festa, S. Valente, R., Piciucchi, M., Panzuto, F., Jensen, R., & Delle Fave, G. (2012). Molecular pathology and genetics of pancreatic endocrine tumours. Journal of Molecular Endocrinology, 49, R37□R50.
- Castori, M., Morrone, A., Kanitakis, J., & Grammatico, P. (2012). Genetic skin diseases predisposing to basal cell carcinoma. European Journal of Dermatology. 2012 May□Jun;22(3):299□309.
- Crino, P.B., Nathanson, K.L., Henske, E.P. (2006). The tuberous sclerosis complex. New England Journal of Medicine, 2006, 355, 1345□1356.

Literatuurlijst (2)

- Darling, T.N., Skarulis, M.C., Steinberg, S.M., Marx, S.J., Spiegel, A.M., & Turner, M. (1997). Multiple facial angiofibromas and collagenomas in patients with multiple endocrine neoplasia type 1. Archives of Dermatology, 133, 853□857.
- Dourmishev, L. A. & Draganov, P.V. (2009). Paraneoplastic dermatological manifestataion of gastrointestinal malignancies. World Journal of Gastroenterology, 15, 4372□4379.
- Ehst, B., Minzer□Conzetti, K., Swerdlin, A., & Devere, T. (2010). Cutaneous manifestations of internal malignancy. Current Problems in Surgery, 47, 384□445.
- Gammon, A., Jasperson, K., Kohlmann, W., & Burt, R.W. (2009). Hamartomatous polyposis syndromes. Best Practices in Research. Clinical Gastroenterology, 23, 219□231.
- Gardner, E.J. (1962). Follow□up study of a family group exhibiting dominant inheritance for a syndrome including intestinal polyps, osteomas, fibromas and epidermal cysts. Am J Hum Genet, 14, 376.

Literatuurlijst (3)

- Gerstenblith, M.R., Goldstein, A.M., & Tucker, M.A. (2010). Hereditary genodermatoses with cancer predisposition. Hematology Oncology Clinics of North America, 24, 885□906.
- Hearle, N., Schumacher, V., Menko, Olschwang. S., Boardman. L.A., Gille, J.J.,…Houlston RS. (2006),. Frequency and spectrum of cancers in Peutz□Jeghers syndrome. Clinical Cancer Research, 12, 3209□3215.
- Hildenbrand, C., Burgdorf, W.H. & Lautenschlager, S. (2001). Cowden syndrome—diagnostic skin signs. Dermatology, 202, 362□366.
- Horvath, A., & Stratakis, C.A. (2009). Carney complex and lentigines. Pigment Cell Melanoma Research, 22, 580□587.
- Jones, E.L., Gajdos ,C., & Wani, S. (2012). Peutz□Jeghers: A hamartomatous polyposis syndrome. Clinical Gastroenterolog and Hepatology. Dec 4.
- Juhn, E.,& Khachemoune, A. (2010). Gardner syndrome: Skin manifestations, differential diagnosis and management. American Journal of Clinical Dermatology, 11, 117□122.

Literatuurlijst (4)

- Karalis, A., Tischkowitz, M., & Millington, G.W. (2011). Dermatological manifestations of inherited cancer syndromes in children. British Journal of Dermatology, 164, 245□56.
- Kleyn, C.E., Lai□Cheong, J.E., & Bell, H.K. (2006). Cutaneous manifestations of internal malignancy: Diagnosis and management. American Journal of Clinical Dermatology, 7, 71□84.
- Kotrulja, L., Ozanić□Bulić, S., Sjerobabski□Masnec, I., Situm, M., & Poduje, S. (2010). Photosensitivity skin disorders in childhood. Collegium Antropologicum, 34, S263□S266.
- Lee, M.J., Chung, K.H., Park, J.S., et al. (2011). Multiple endocrine neoplasia type 2B: Early diagnosis by multiple mucosal neuroma and its DNA analysis. Annals of Dermatology, 22, 452□455.
- Lodish, M.B. & Statakis, C.A. (2010). Endocrine tumors in neurofibromatosis type 1, tuberous sclerosis and related syndromes. Best Practices & Research. Clinical Endocrinology & Metabalism, 24, 439□449.

Literatuurlijst (5)

- Lloyd, K.M. Dennis, M. (1963). Cowden's disease. A possible new symptom complex with multiple system involvement. Annals of Internal Medicine, 58, 136□142.
- Lookingbill et al. Skin involvement as the presenting sign of internal carcinoma. *J Am Acad Dermatol* 22:19-26, 1990
- Mann, J.A. & Siegel, D.H. (2009). Common genodermatoses: What the pediatrician needs to know. Pediatric Annals, 38, 91□98.
- Muir, E. G., Bell, A.J., & Barlow, K.A. (1967). Multiple primary carcinomata of the colon, duodenum, and larynx associated with keratoacanthomata of the face. British Journal of Surgery, 54, 191□195.
- Schrager, C.A., Schneider, D., Gruener, A.C., et al. (1998). Clinical and pathological features of breast disease in Cowden's syndrome: an underrecognized syndrome with an increased risk of breast cancer. Human Pathology, 29, 47□53.

Literatuurlijst (6)

- Okan, G., Vural, P, Ince, U., Yazar, A., Uras, C., & Saruc, M. (2012). Muir□Torre syndrome: A case report and review of the literature. Turkish Journal of Gastroenterology, 23, 394□398.
- Parks, E.T., Caldemeyer, K.S., Mirowski, G.W. (2001). Gardner syndrome. Journal of the American Academy of Dermatology, 45, 940□942.
- Pilarski, R.(2009). Cowden syndrome: A critical review of the clinical literature. Journal of Genetic Counseling, 18, 13□27
- Roach, E.S., Gomez, M.R., & Northrup, H. (1998). Tuberous Sclerosis Consensus conference: Revised clinical diagnostic criteria. J Child Neurology, 13, 624□628.
- Sabir et al. Cutaneous manifestations of cancer. *Curr Opin Oncol* 11:139-44,1999.
- Mukamal, L.V., Ferreira, A.F., de Moura astro Jacques, C., Amorim, C.A.F.L., & Pineiro□Maceira, J. (2012). Cowden syndrome; Review and report of a case of late diagnosis. International Journal of Dermatology, 51, 1494□1499.

Literatuurlijst (7)

- Schwartz. Acanthosis nigricans. *J Am Acad Dermatol* 31:1-19, 1994
- Shen, Z., Hoffman, J.D., Hao, F., & Pier, E. (2012). More than just skin deep: Faciocutanoeus clues to genetic syndromes with malignancies. Oncologist, 17, 930□936.
- Sigurgeisson et al. Risk of cancer in patients with dermatomyositis or polymyositis. *N Engl J Med* 326:363-7, 1992
- Torre, D. (1968). Multiple sebaceous tumors. Archives of Dermatology, 98, 549□551.
- Vezzosi, D., Vignaux, O., Dupin, N., & Bertherat, J. (2010). Carney complex: Clinical and genetic 2010 update. Annals of Endocrinology, 71, 486□493.
- Winship, I.M., Dudding, T.E. (2008). Lessons from the skin—cutaneous features of familial cancer. Lancet Oncology, 9, 462□472