
Good Morning
YEAH!
SaraL

Fascinating **F**eatures and **M**ysterious **M**anifestations of Disease

OLLI @ University of Illinois

Fall 2022 Semester

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Session 4

Diseases with Unusual Appearance

October 7, 2022

Plan for the Course

- Session 1: Diseases with a color
- Session 2: Diseases with an odor or a taste
- Session 3: Textures or Sounds of Disease
- Session 4: Diseases with unusual appearance
- Session 5: Flying, crawling & burrowing critters
- Session 6: Forgotten or ignored epidemics
- Session 7: } Medical detectives solve mysterious cases
- Session 8: }

Plan for the Session

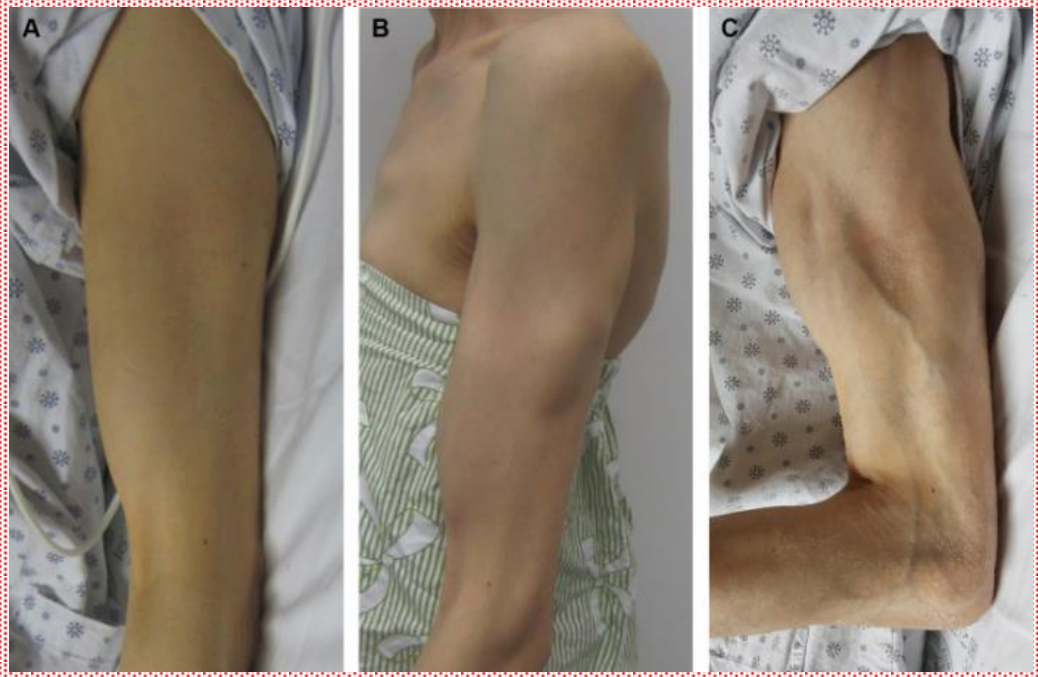
- Stiff Person Syndrome (SPS)
- Gigantism and Acromegaly
- Hypertrichosis & Uncombable Hair Syndrome
- Dermatographia (Skin writing)
- Proteus Syndrome and Moebius Syndrome
- Leprosy
- Ichthyosis, Alligator Skin, Harlequin skin
- Carcinoid Syndrome
- Ochoa Syndrome
- Situs Inversus Totalis (SIT)

STIFF PERSON SYNDROME (SPS)

Stiff-Person Syndrome (SPS)

- A rare progressive autoimmune movement disorder of the CNS that causes muscle rigidity in the limbs (also known as Moersch-Woltman syndrome).
- Accompanied by heightened sensitivity, with muscle spasms triggered by noise, emotional distress or light physical touch.
- Patients first experience stiffening of the trunk muscles, followed by stiffness and rigidity in the legs and other muscles in the body.

Stiff-Person Syndrome (SPS)



Patients may suffer from:

- Disabled movement
- Abnormal postures
- Difficulty functioning in busy environments
- Spasms so strong they can cause bone fractures
- Misconnections in the brain or spinal cord

Incidence:

- About 1 in 1M people have this diagnosis.
- Twice as frequent in women than in men.
- Any age but usually between ages 30 and 60.

Stiff-Person Syndrome (SPS)

Predisposing factors

Autoimmune disorders

- Diabetes
- Thyroiditis
- Vitiligo
- Pernicious anemia.

Malignancies

- Breast
- Lung
- Kidney
- Thyroid
- Colon
- Hodgkin’s lymphoma.

Stiff-Person Syndrome (SPS)

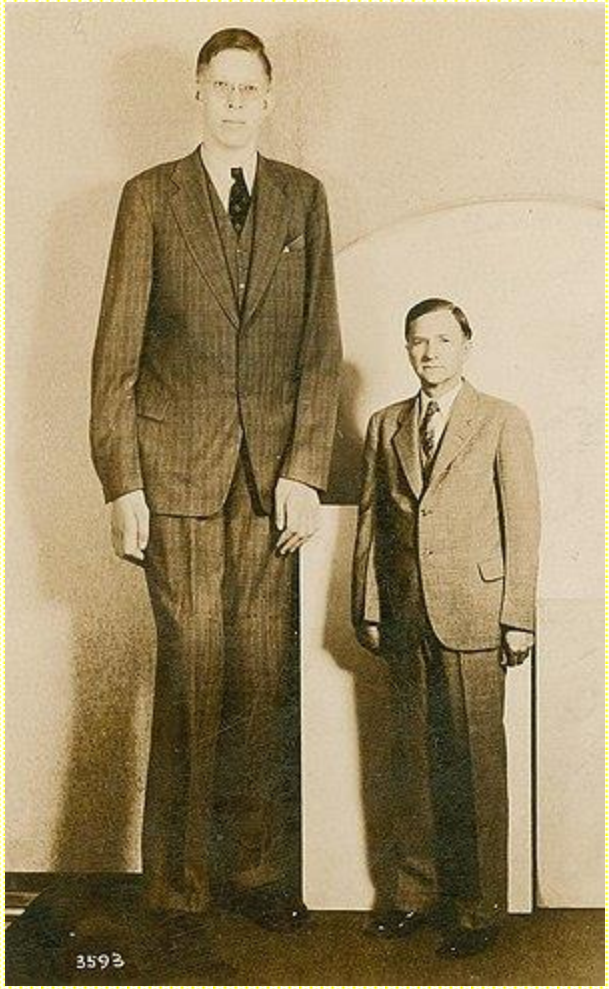
- People with this disorder make antibodies that attack an enzyme called *glutamic acid decarboxylase* (GAD).
- GAD makes a neurotransmitter which helps control muscle movement called *gamma-aminobutyric acid* (GABA).
- Researchers believe that the immune system in people with SPS mistakenly attacks GAD enzyme, which lowers the amount of GABA in the body.

Stiff-Person Syndrome (SPS)

Treatment

- Oral meds include benzodiazepines or baclofen for muscle stiffness and spasms, and anti-seizure drugs to lessen pain.
- Occasional use of anti-inflammatories and corticosteroids may be useful in some cases for pain flares.
- IV meds include immunoglobulin (IVIG), plasmapheresis, rituximab and autologous stem cell transplant.
- Non-medication options include physical therapy, massage, water therapy, heat therapy, acupuncture and others.

Robert Wadlow 8’ 11”



GIGANTISM

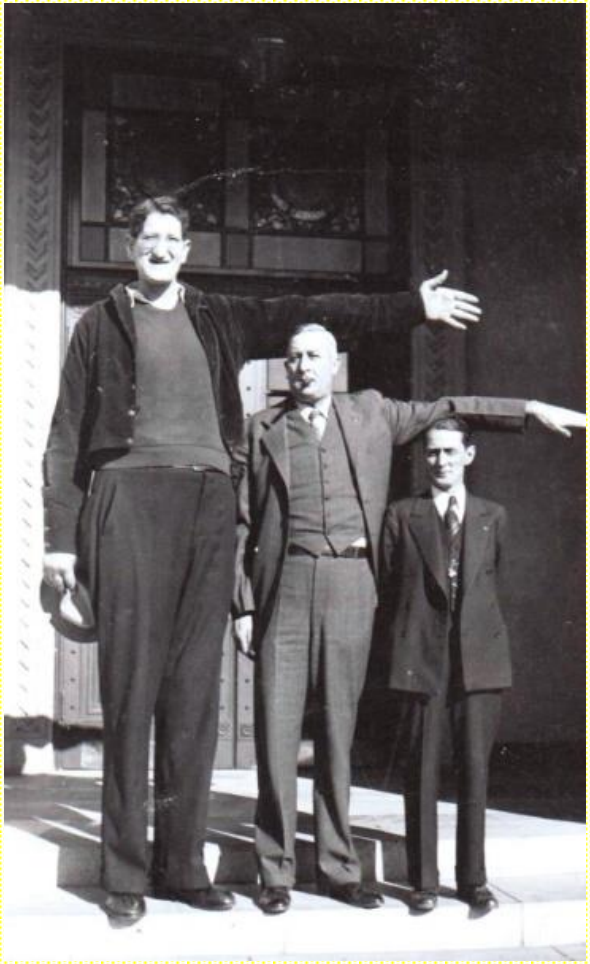
Gigantism

- Gigantism is an incredibly rare growth hormone (*hGH*) disease that causes a person to grow abnormally tall.
- A benign tumor (adenoma) in the pituitary gland overproduces the *hGH* during pre-adolescence.
- The disease is treatable and the overgrowth of bones, can be stopped.
- If left untreated, people can get to be over 8 ft. tall.

Gigantism

- Increased levels of *hGH* in childhood before the fusion of the bone growth plates at puberty.
- Excessive growth with height significantly above average: people about 2 - 3 m (8 - 9 ft.) in height.
- Should not be confused with *acromegaly*, which involves adult somatic enlargement in the face and extremities.

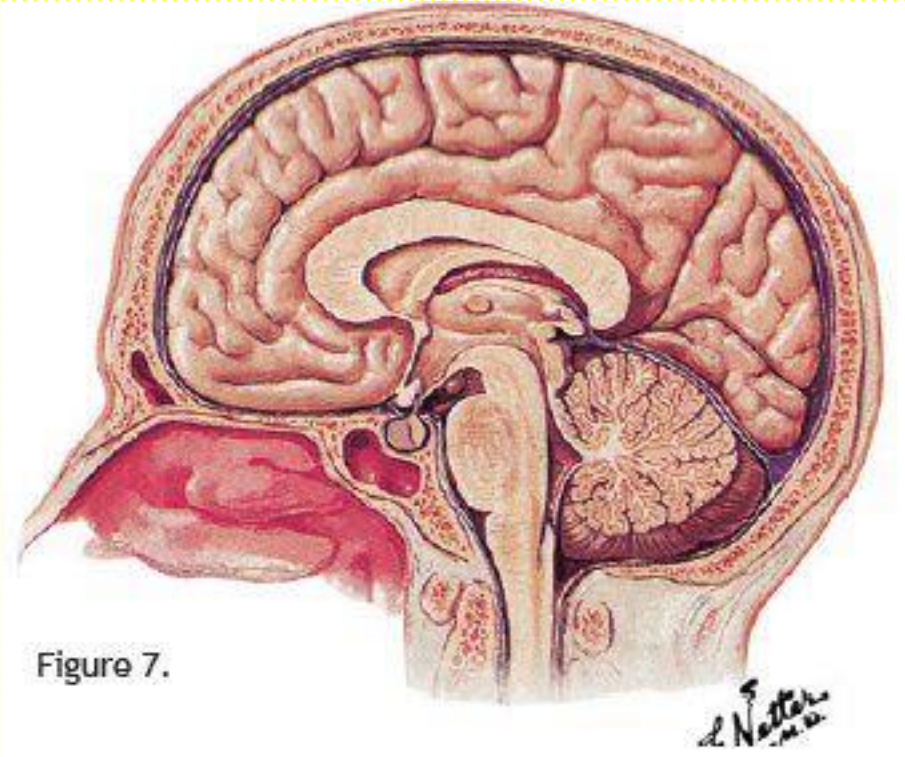
Gigantism



Jake Erlich 8’ 6”

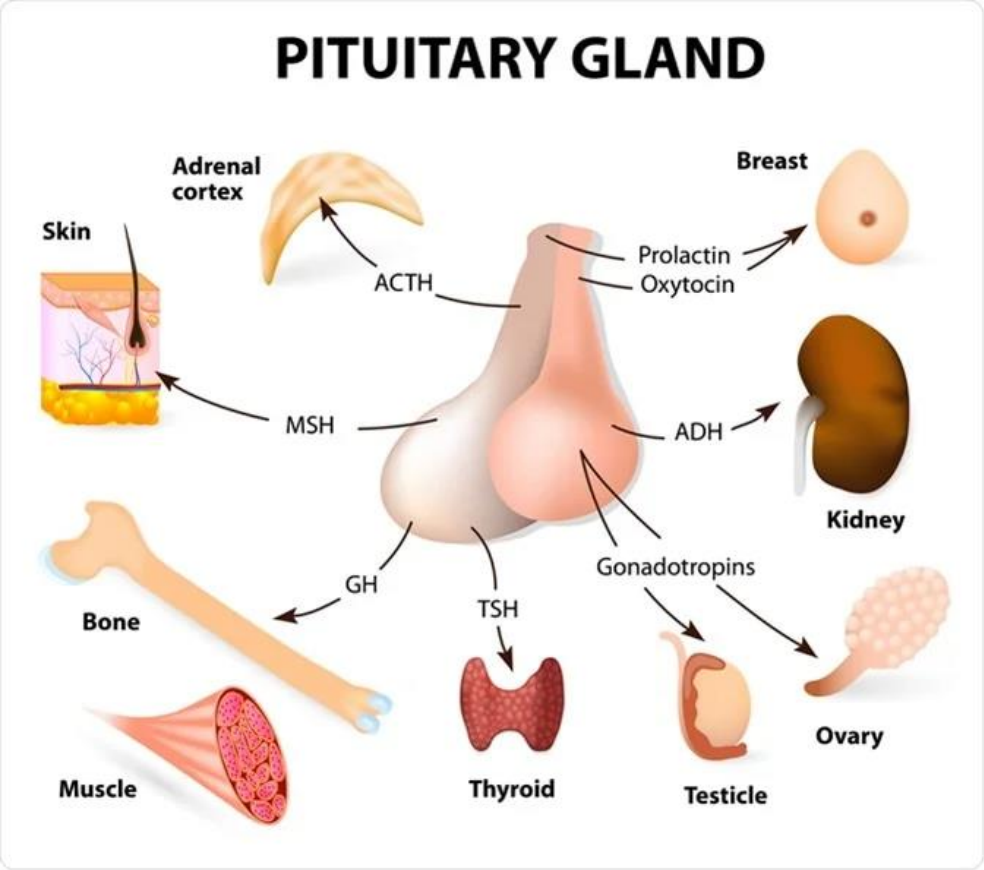
- Several gene mutations have been associated with gigantism, but over 50 % of cases cannot be linked to genetic causes.
- People with gigantism have multiple health problems involving the circulatory or skeletal systems, as there are abnormal demands on both the bones and the heart.

The Pituitary gland



Sella Turcica (“Turkish Saddle”)

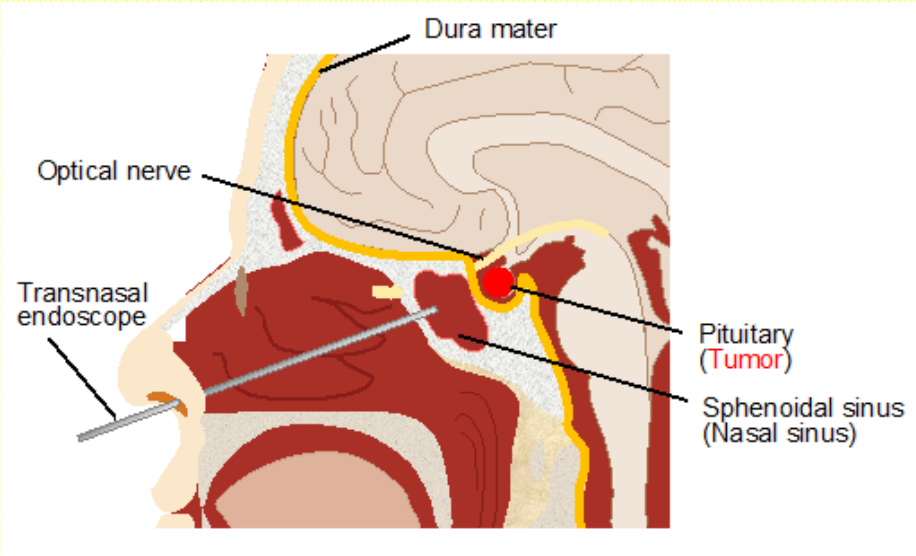
Pituitary (Hypophysis)



Pituitary Hormones
Adrenocorticotrophic (ACTH)
Prolactin (PRL)
Oxytocin (hypothalamus)
Anti-diuretic (ADH)
Luteinizing (LH)
Follicle-stimulating (FSH)
Thyroid-stimulating (TSH)
Growth (GH)
Melanocyte-stimulating (MSH)

Gigantism

- ***Pegvisomant*** can significantly decrease high growth rates if paired with radiation.
- Surgery for the adenoma can sometimes be done.
- Resulting from surgery and/or radiation therapy, about 60% of people with gigantism develop ***hypo****pituitarism* after treatment.
- Lack of one, several or all the hormones made by the pituitary gland requires hormone replacement therapy.



ACROMEGALY

Acromegaly

- Disorder resulting from excess growth hormone (hGH) after the growth plates have closed.
- Starts with enlargement of the hands and feet, and also of the forehead, jaw, and nose.
- Other symptoms may include joint pain, thicker skin, deepening of the voice, headaches, and vision problems.
- Complications may include type 2 diabetes, sleep apnea, and high blood pressure.

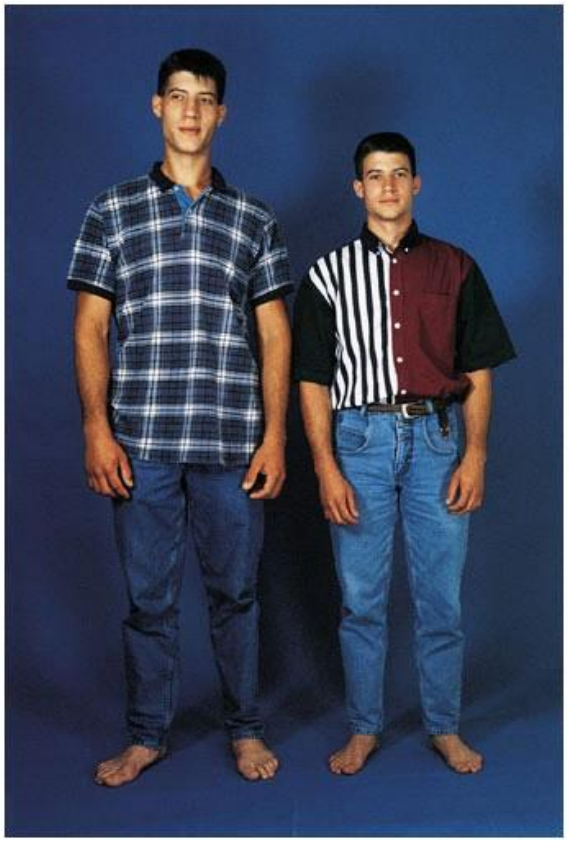
Acromegaly

- 95% of cases caused by a pituitary adenoma.
- Affects about 3 per 50,000 people.
- Most commonly diagnosed in middle age.
- Males and females are affected with equal frequency.

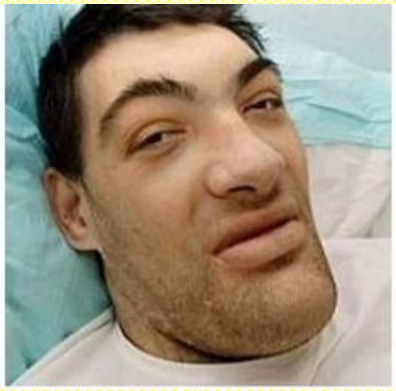
Acromegaly

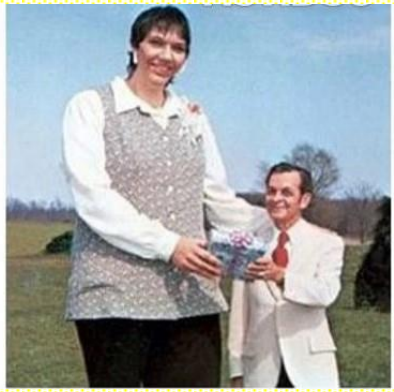
- From the Greek: "*akron*", extremity, and "*mega*", large.
- Adenoma can cause visual problems, headaches, and problems with other hormones.
- Rarely, acromegaly is caused by tumors of the pancreas, lungs, and adrenal glands and not the pituitary.
- These tumors lead to an excess of *hGH*, because they produce it, or, more frequently, because they produce *GHRH* (growth hormone-releasing hormone).
- When these non-pituitary tumors are surgically removed, *hGH* levels fall and the symptoms of acromegaly improve.

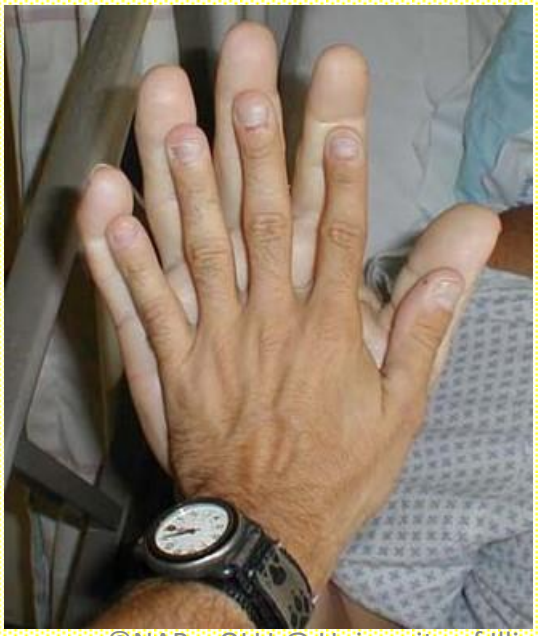
Acromegaly

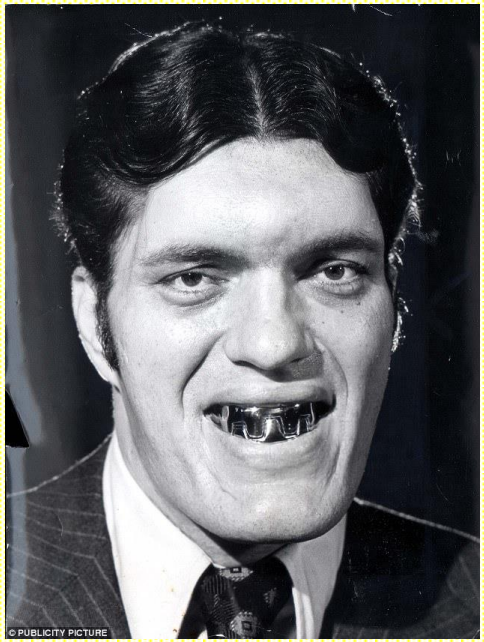


Twins









HYPERTRICHOSIS
(WEREFOLF MAN)

Werewolf Man

(Hypertrichosis)

- Extreme hair growth on the patient's body.
- Extremely rare, it can occur at any stage of life.
- Different forms of the disease that can affect patients differently.
- Not only men, and can cover anywhere from head to toe, in small or big patches.

Werewolf Man
(Hypertrichosis)

Petrus Gonsalvus
(Pedro González)

Madelene Gonsalvus
1580

Antoinetta Gonsalvus
1583

Henry Gonsalvus
1585

Petrus and his wife Catherine
1595

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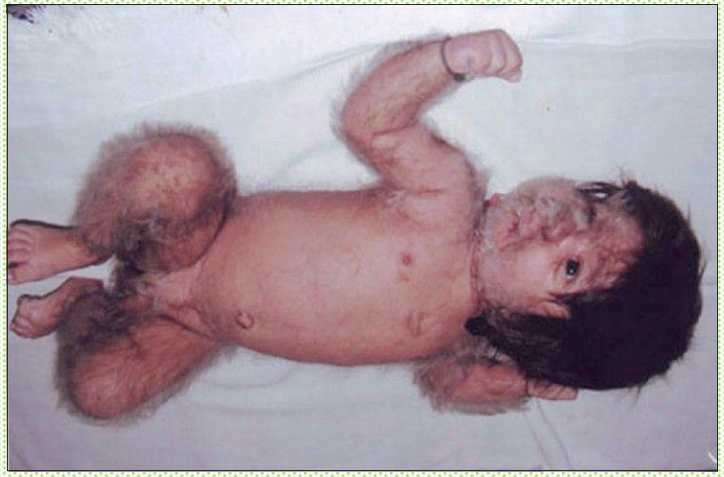
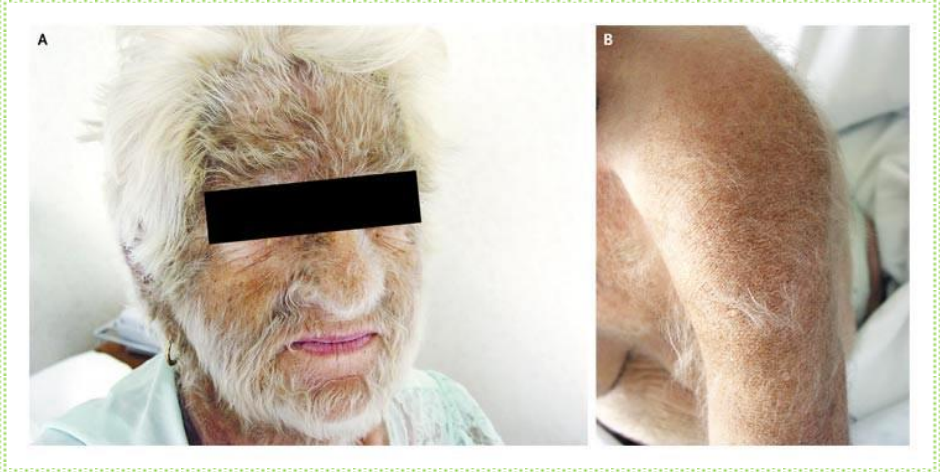
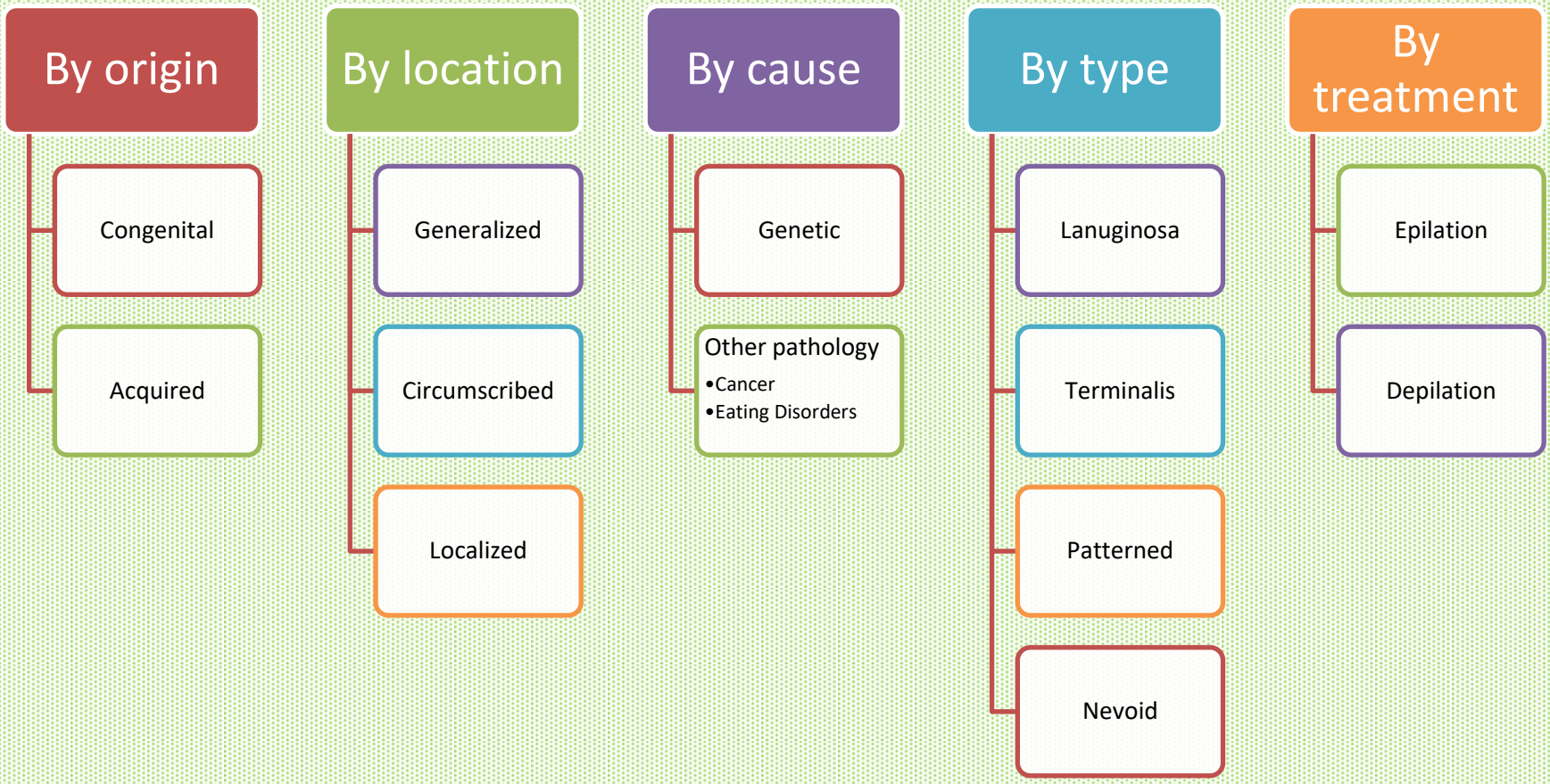
Werewolf Man (Hypertrichosis)





Werewolf Man
(Hypertrichosis)



Ramírez 2022

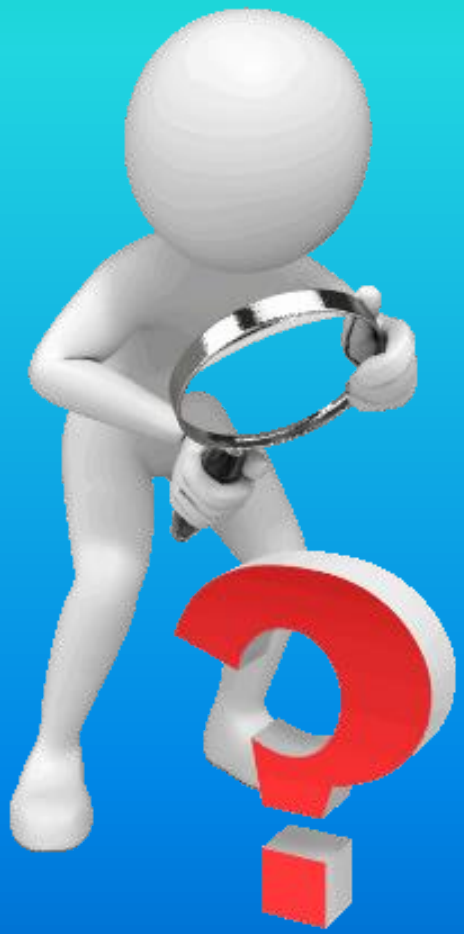
Werewolf Man

(Hypertrichosis)

- Victor "Larry" Gómez, Gabriel "Danny" Ramos Gómez, Luisa Lilia De Lira Aceves and Jesús Manuel Fajardo Aceves are 4 of a family of 19 that span 5 generations suffering from Congenital Generalized Hypertrichosis.

- Women are covered with a light-to-medium coat of hair while men have thick hair on about 98% of their body excepting hands and feet.

Questions? 1



UNCOMBABLE HAIR SYNDROME (UHS)

Uncombable Hair Syndrome (UHS)

Extremely rare condition (100 confirmed cases):

- It develops as early as infancy and usually before age 3, but can develop as late as 12.
- It is characterized by highly frizzy, matted, and dry hair that cannot be combed flat.
- It occurs only on the scalp, and grows in every direction.
- It can improve over time, but usually causes discomfort for the individual growing up.

Uncombable Hair Syndrome (UHS)

- Disorder of the hair shaft of the scalp, with silvery blond or straw-colored hair that is disorderly and cannot be combed flat.
- It has been described in association with other diseases, such as ectodermal dysplasias.
- Despite its appearance, the hair is not fragile or brittle, and it grows at a normal or slightly slower rate.

Uncombable Hair Syndrome (UHS)

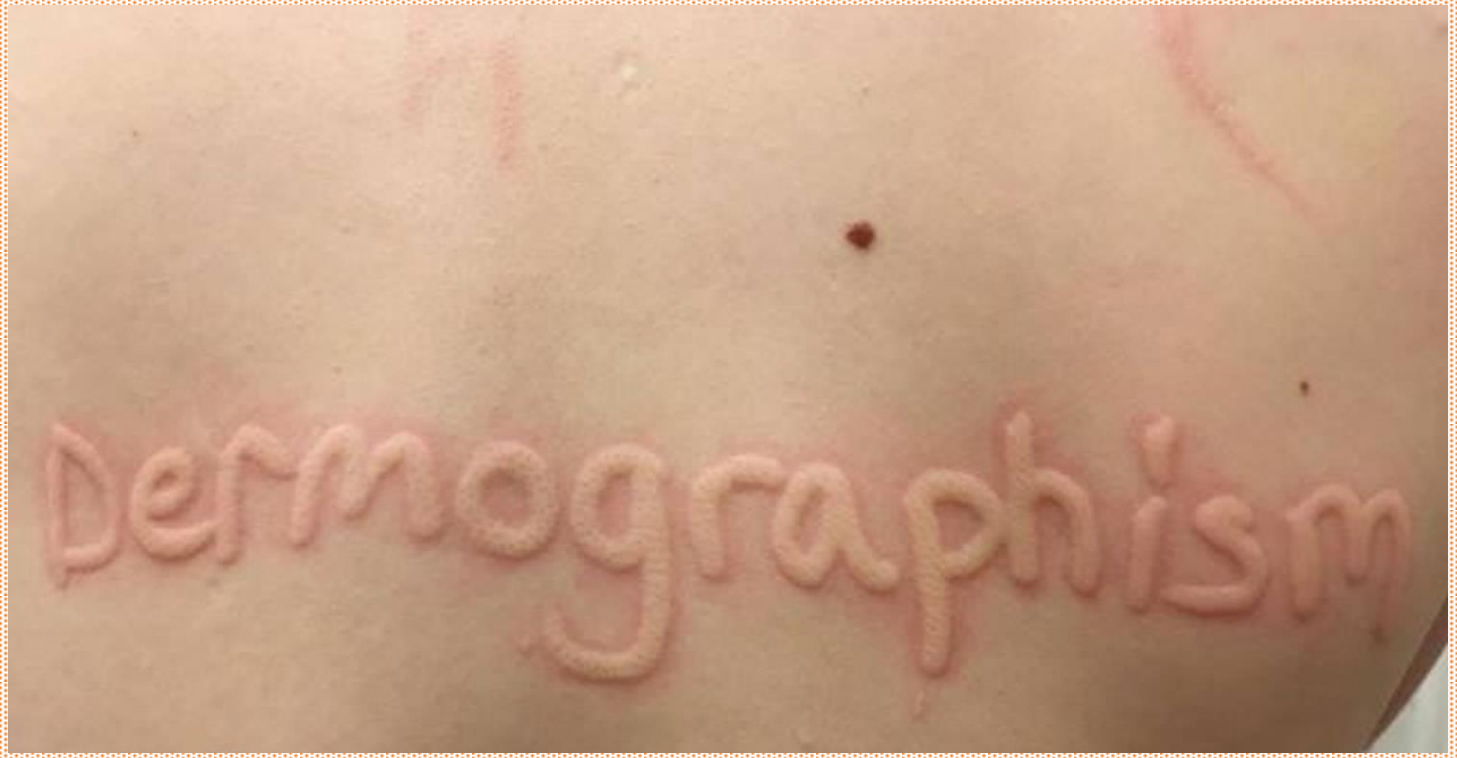
- Caused by genetic changes in 3 genes that code for proteins involved in hair shaft formation.
- Autosomal recessive but cases inherited in an autosomal dominant manner may also exist.
- Some research per the Genetic And Rare Disease (GARD) Information Center has found that biotin (B7) supplements may improve the condition.

Uncombable Hair Syndrome (UHS)

- Other names:
  - Spun glass hair.
  - Cheveux incoiffables.
  - Pili trianguli et canaliculi.

- Hair shafts, normally round to oval in cross-section, may have triangular cross-sections or grooves along their length which do not allow the hairs to lie flat.

Uncombable Hair Syndrome (UHS)



SKIN WRITING
(DERMATOGRAPHIA)

Skin Writing

Dermatographia

- Writing or drawings can be made to appear on skin by stroking it with a fingernail or a closed ball-point pen.
- Initially, skin will turn white, then the redness and swelling start, taking the form of the strokes that were made.
- Cause is unknown, but may be related to allergies or episodes of emotional stress.
- Treated with antihistamines like Zyrtec and Benadryl.

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Skin Writing

Dermatographia

Symptoms:

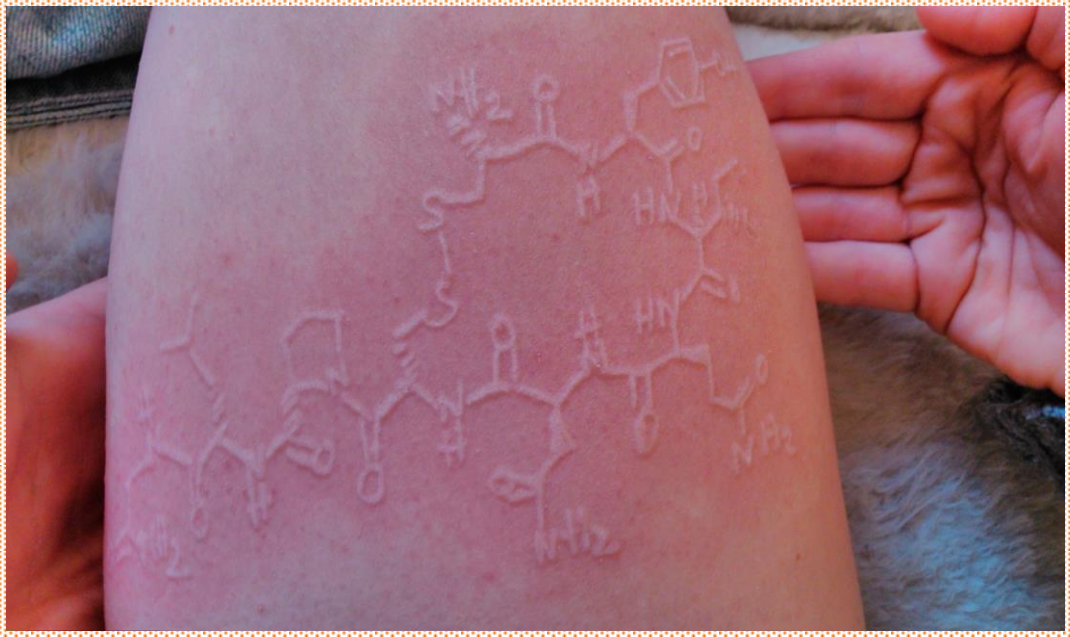
- Raised red welts where skin was scratched or irritated
- Welts or hives develop within 5 to 7 minutes of scratching
- Itching
- Usually fade away within 15 minutes to ½hour.
- Rarely, symptoms may develop more slowly and take hours or days to disappear.

Skin Writing

Dermatographia

Triggers

Excessively dry skin.
Exercise.
Strong emotions or stress.
Cold weather.
Hot water.
Infections: viruses, bacteria, fungi.
Certain medications, like penicillin.



Risk Factors

Thyroid conditions.
Diabetes.
Reactive polyarthritis.
Behcet's disease.
Pregnancy.
Menopause.

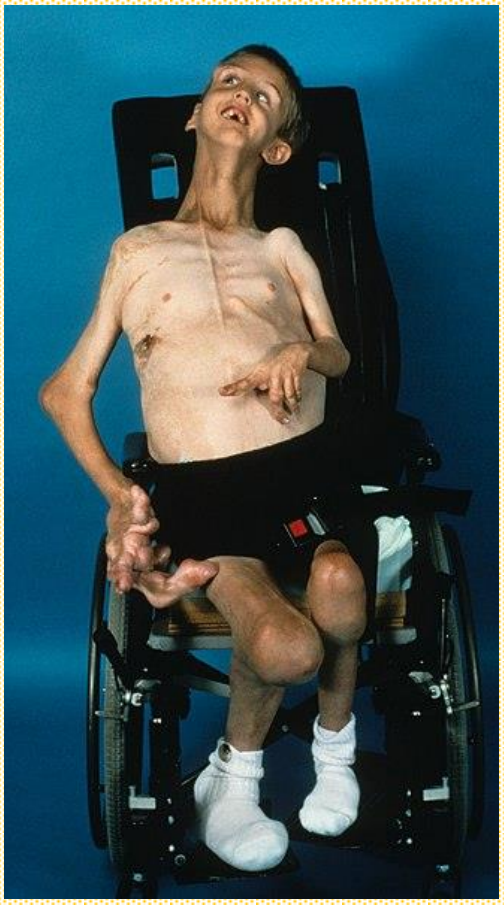
The Good News

Not considered dangerous.
Doesn’t cause any health issues.
In 33% of people it goes away in 5 years.
In many people, lasts as little as 10 days.

Skin Writing

Dermatographia

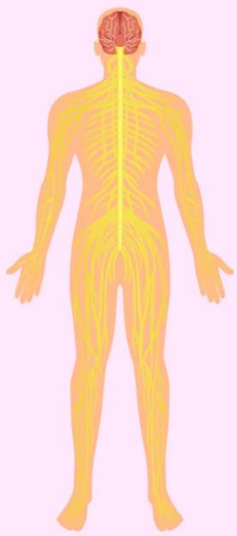
- 2% to 5% of people have dermatographia.
- It is a type of allergic reaction.
- In 40%, severe itching interferes with daily life.
- Younger people, like children, more affected.



PROTEUS SYNDROME

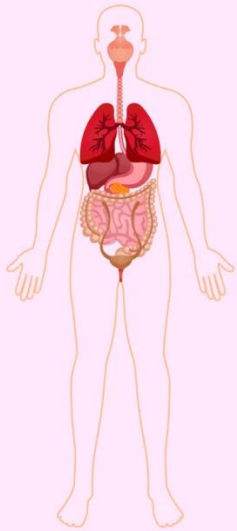
Embryogenesis

Embryo beginning organogenesis



Ectoderm

Mesoderm



Endoderm

CNS and superficial tissues like the epidermis

Muscular, skeletal, cartilage, urogenital, and blood systems

Internal organs of the digestive and respiratory systems.

Proteus Syndrome

- A peculiar overgrowth in bones, skin, or other tissues, which makes that part of the body grow out of proportion.
- All three embryonic cell layers can be affected (*endoderm, mesoderm* and *ectoderm*).
- The inflammation of connective tissue can lead to serious impairments and permanent, abnormal physical and facial features.

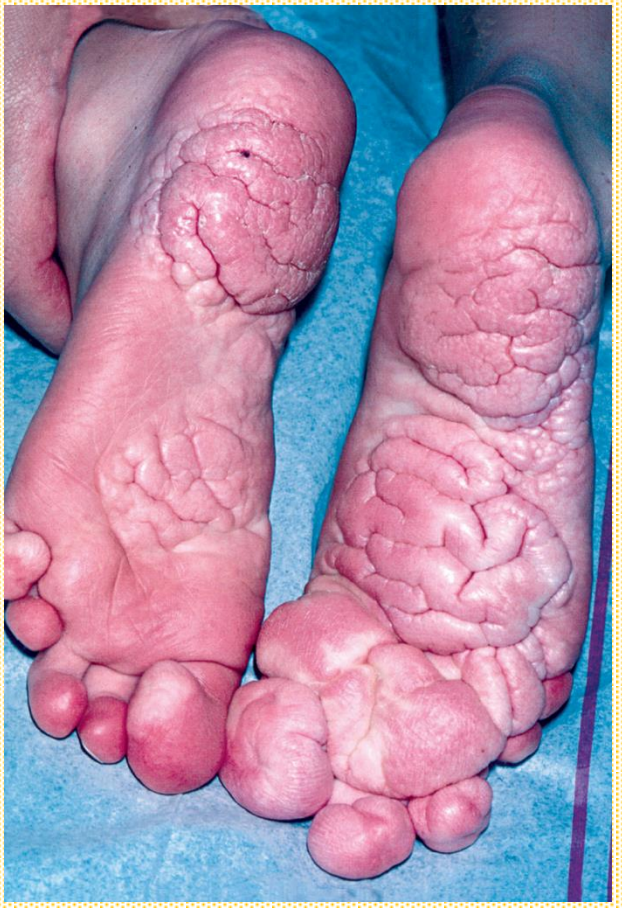
Proteus Syndrome

Symptoms

- Asymmetric overgrowths.
- Raised, rough skin lesions with grooved appearance
- A curved spine (*scoliosis*).
- Fatty overgrowths on the face, arms, and legs.
- Noncancerous tumors, on ovaries and meninges.
- Mental disabilities.
- Malformed blood vessels, life-threatening clots.
- Malformation of the central nervous system.
- Long face, narrow head, droopy eyelids, and wide nostrils.
- *Cerebroid nevus* on the soles of the feet.

Proteus Syndrome

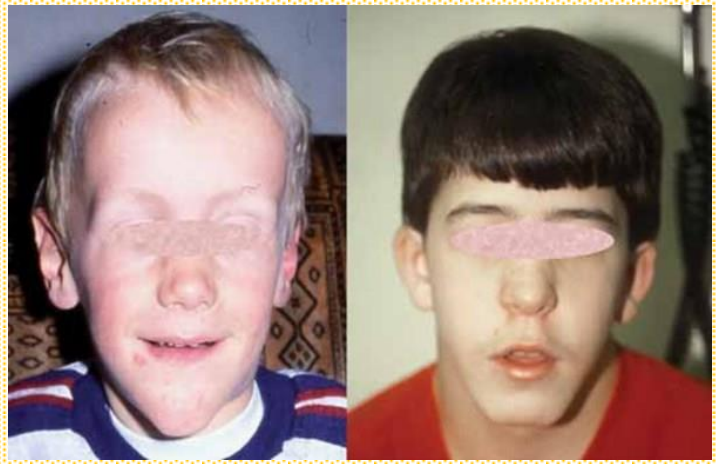
- Newborns with Proteus syndrome have few or no signs of the condition.
- Overgrowth becomes apparent between the ages of 6 and 18 months and gets more severe with age.
- A thick, raised, and deeply grooved skin lesion known as a *cerebriform* nevus appears on soles.
- Blood vessel, fat tissue and face also show abnormal growth.

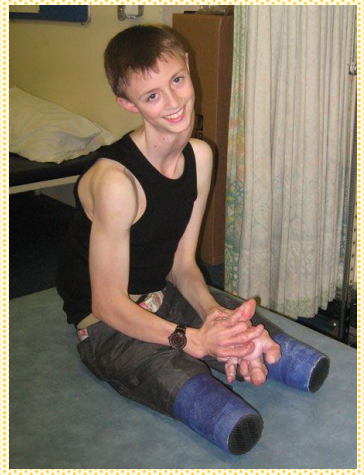


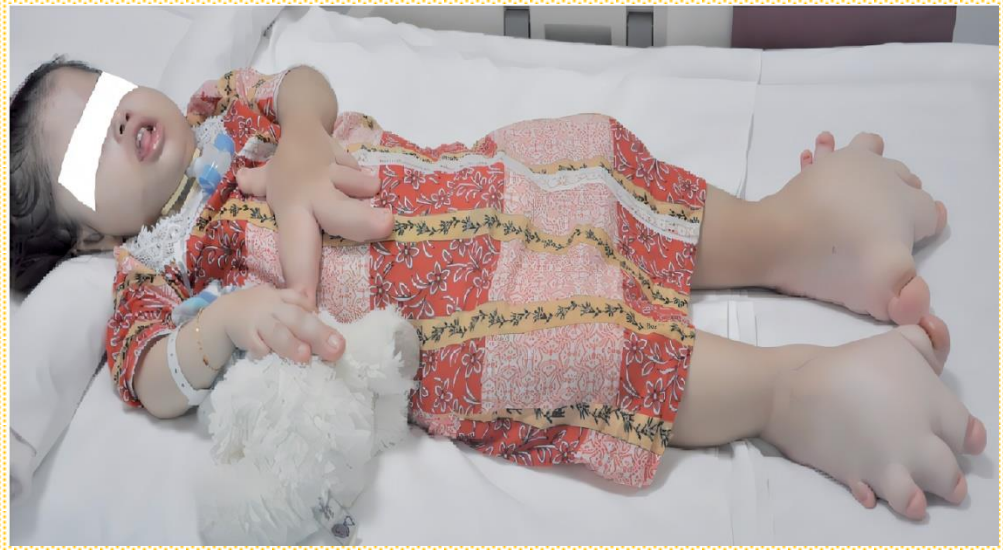
Proteus Syndrome

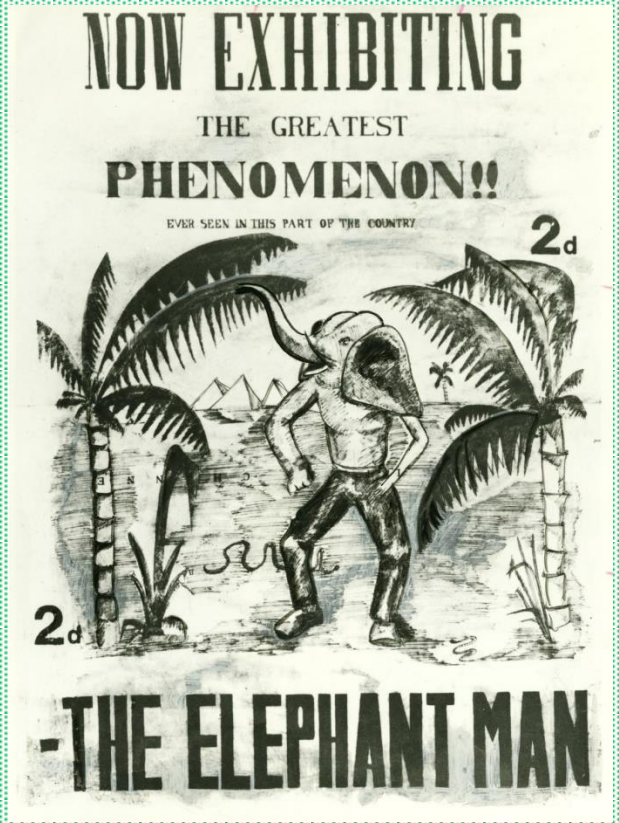
- Some people have neurological abnormalities, including intellectual disability, seizures, and vision loss.
- It is unclear why people with neurological symptoms are more likely to have distinctive facial features.
- It is also unclear how these signs and symptoms are related to abnormal growth.

Proteus Syndrome









Poster for a London appearance

The ELEPHANT MAN

The Elephant Man

Joseph Merrick (1862-1890)

- Became a 'freak show exhibit' in 1884 and was exploited most of his life.
- Mother thought that a fright episode during pregnancy caused her child’s deformities.
- He slept sitting, his legs drawn up and used as a headrest because the weight of his huge head could crush his neck.

The Elephant Man

Joseph Merrick (1862-1890)

- Sir Frederick Treves, a Doctor from London Hospital befriended him, and later obtained for him room and board in the hospital's basement.
- He spent the last 4 years of his life there, and died in 1890, from asphyxia.
- He said those were probably the best years of his life.

The Elephant Man

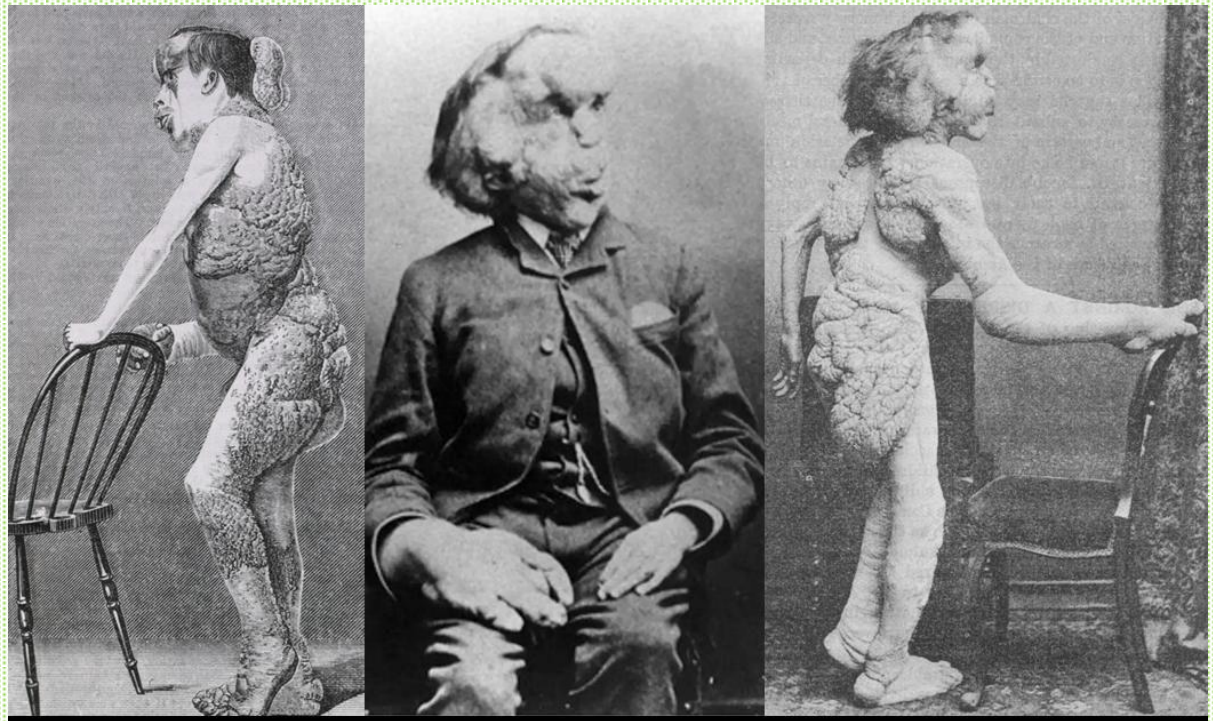
Joseph Merrick (1862-1890)

- Exact diagnosis is still uncertain.
- DNA analysis of hair and bones is inconclusive.
- Most clinicians believed that he had severe neurofibromatosis.
- Nowadays, the thought is that he had Proteus syndrome.

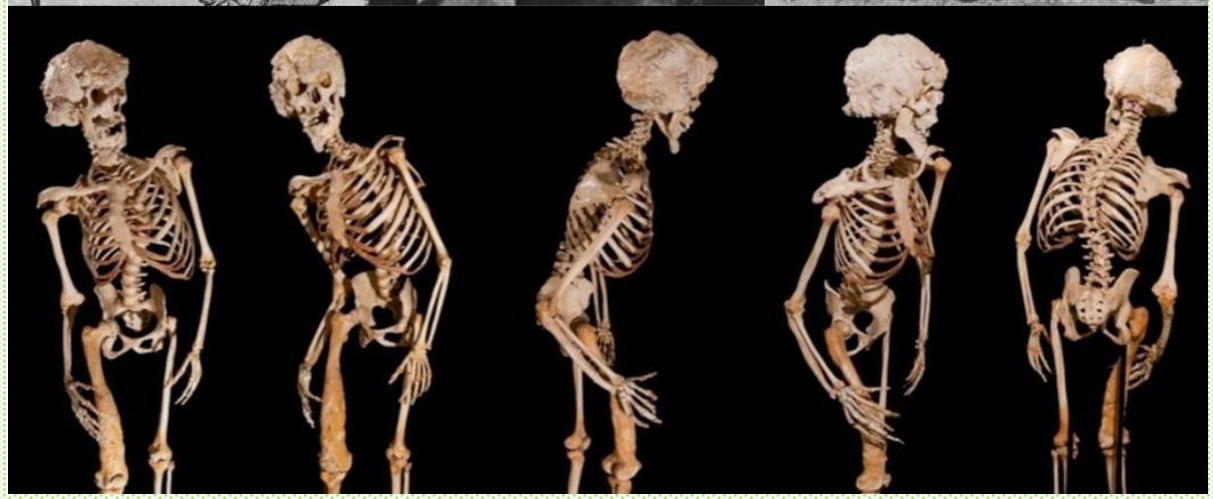
Elephant Man

Joseph Merrick

(1862-1890)



Photographs in 1889



Skeleton In London Hospital

TREE MAN SYNDROME
EPIDERMODYSPLASIA VERRUCIFORMIS (EV)

Tree Man Syndrome

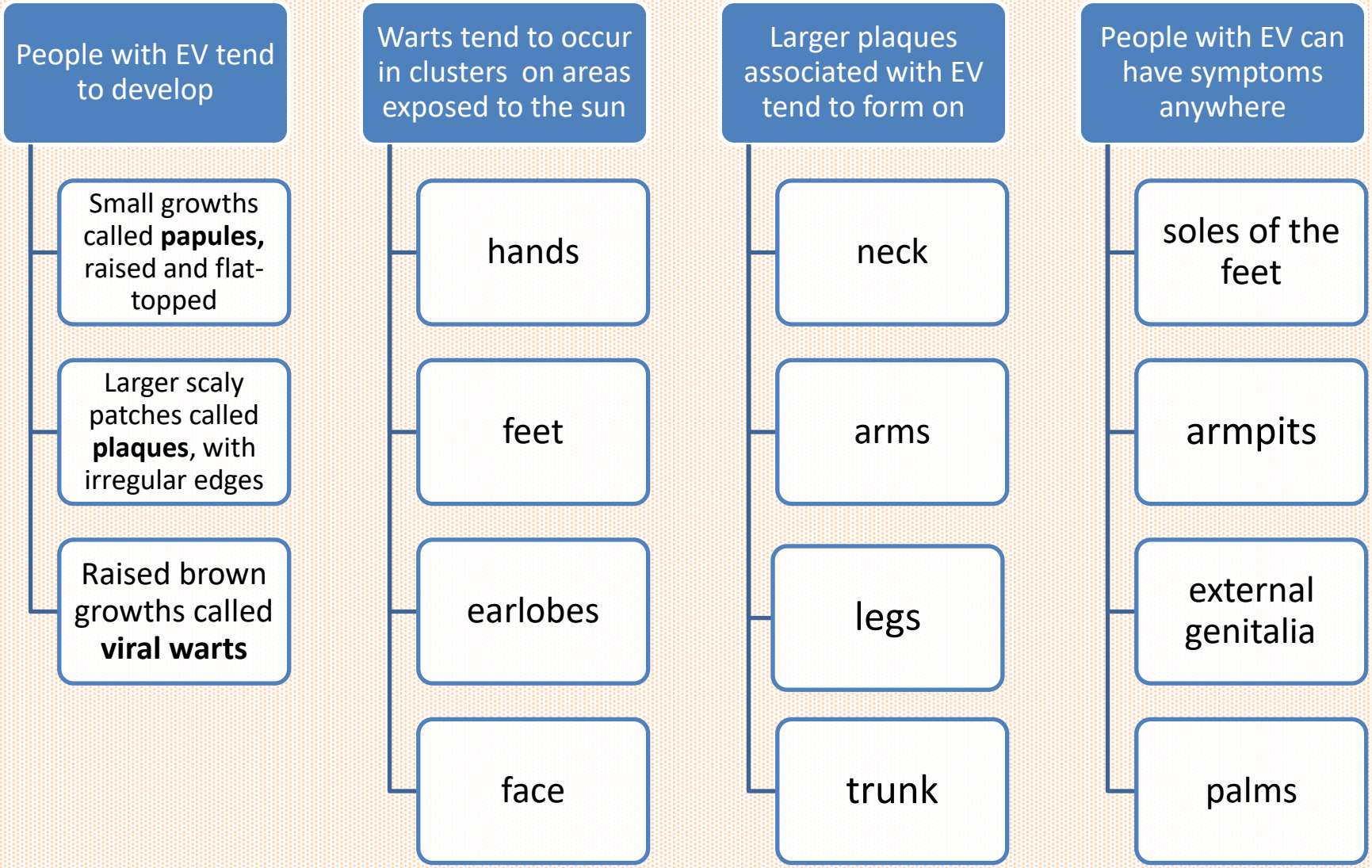
Epidermodysplasia verruciformis (EV)

- This bizarre disease is an incredibly rare *autosomal recessive* hereditary skin condition with a high risk of skin cancer.

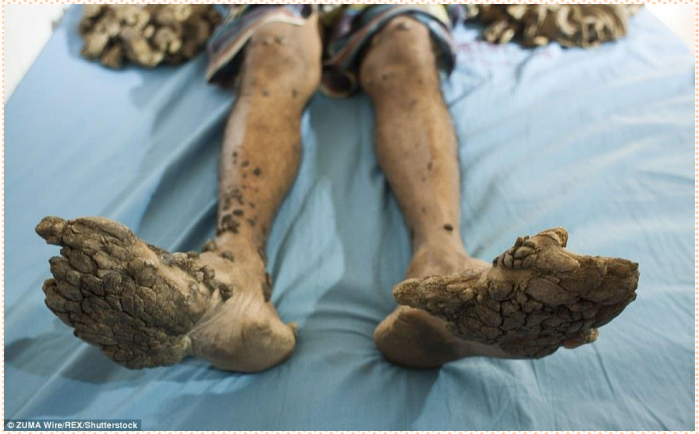
- Abnormal susceptibility to papillomavirus of the skin, which causes scaly or bark-looking growth on the hands and feet.

- Usually diagnosable before the age of 20.

Skin Symptoms in EV



Tree Man Syndrome



Tree Man Syndrome

Epidermodysplasia verruciformis (EV)

- 7.5% of patients develop EV in infancy, 61.5% in childhood, and 22% during puberty.
- In ~10% of EV cases, the person's parents were relatives.
- Lesions continue to develop throughout a person's life.
- Acquired EV has developed in people with: lymphoma, HIV, and organ transplants.

EV and HPV Vaccine

- Seven cancer-causing HPV types: 16, 18, 31, 33, 45, 52, and 58 are targeted by Gardasil.
- Types of HPV typically associated with the development of skin cancer in people with EV are: HPV 5, 8, 17, 20, and 47 — especially types 5 and 8.
- So, vaccine is probably not useful.

Tree Man Syndrome

Therapies

Medication

- Systemic or topical retinoids: prevent spread, decrease inflammation.
- Interferon-alpha: reduces cell growth and division, treats viral infections.
- Cholecalciferol: (D3) modulates inflammatory and immune responses.

Lifestyle changes:

- Sun avoidance or protection: prevent skin cancer.
- Quit smoking: Smoking increases risk of cancer and reduces immune function.

Cryotherapy: Treezing off growths.

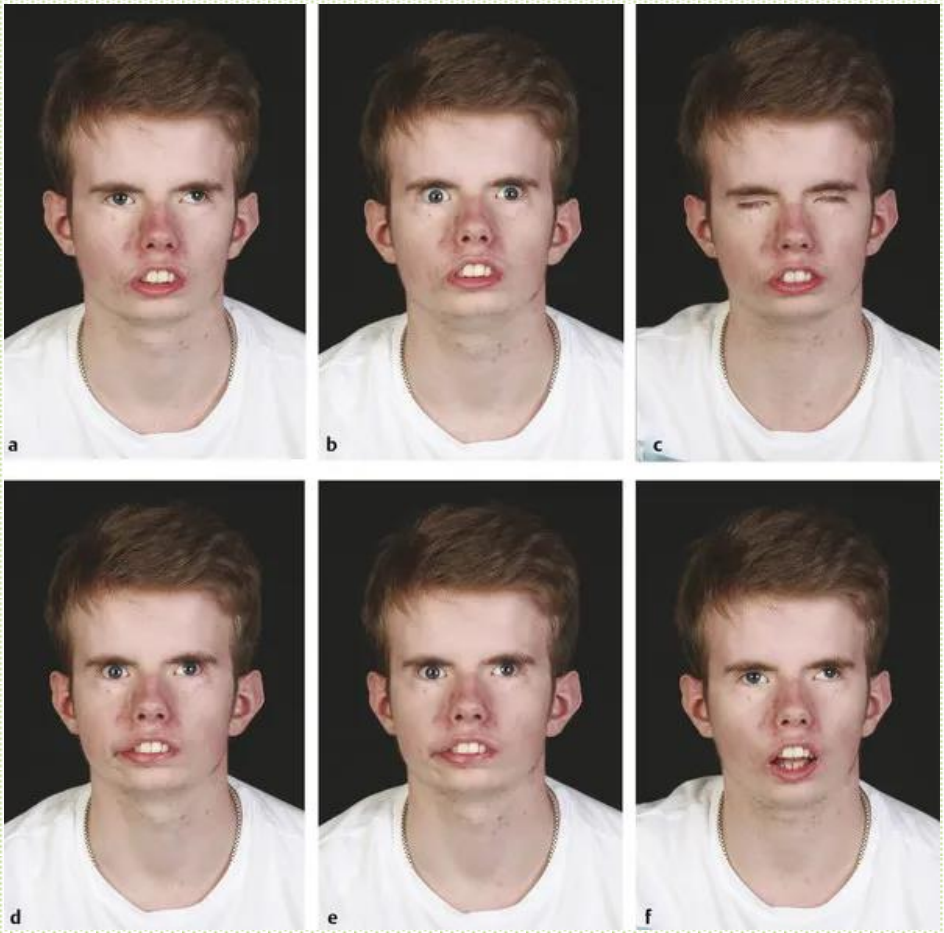
Surgery: Lasers or high-frequency currents to remove or destroy growths.

MOEBIUS SYNDROME

Moebius Syndrome

- Rare congenital condition with underdevelopment or absence on both sides of the face, of the nerves that control some eye movements and facial expressions.
- It can also affect the nerves that control speech, chewing, and swallowing.
- Some cases suggest an increased risk of parent-to-child transmission so expectant families with a Moebius history may benefit from genetic counseling.

Moebius Syndrome



- It's sometimes called "frozen face syndrome".
- Affects muscles that control back-and-forth eye movement, so patients must move their head from side to side to read or follow the movement of objects.
- Eyelids may not close completely when blinking or sleeping, which can result in dry or irritated eyes.

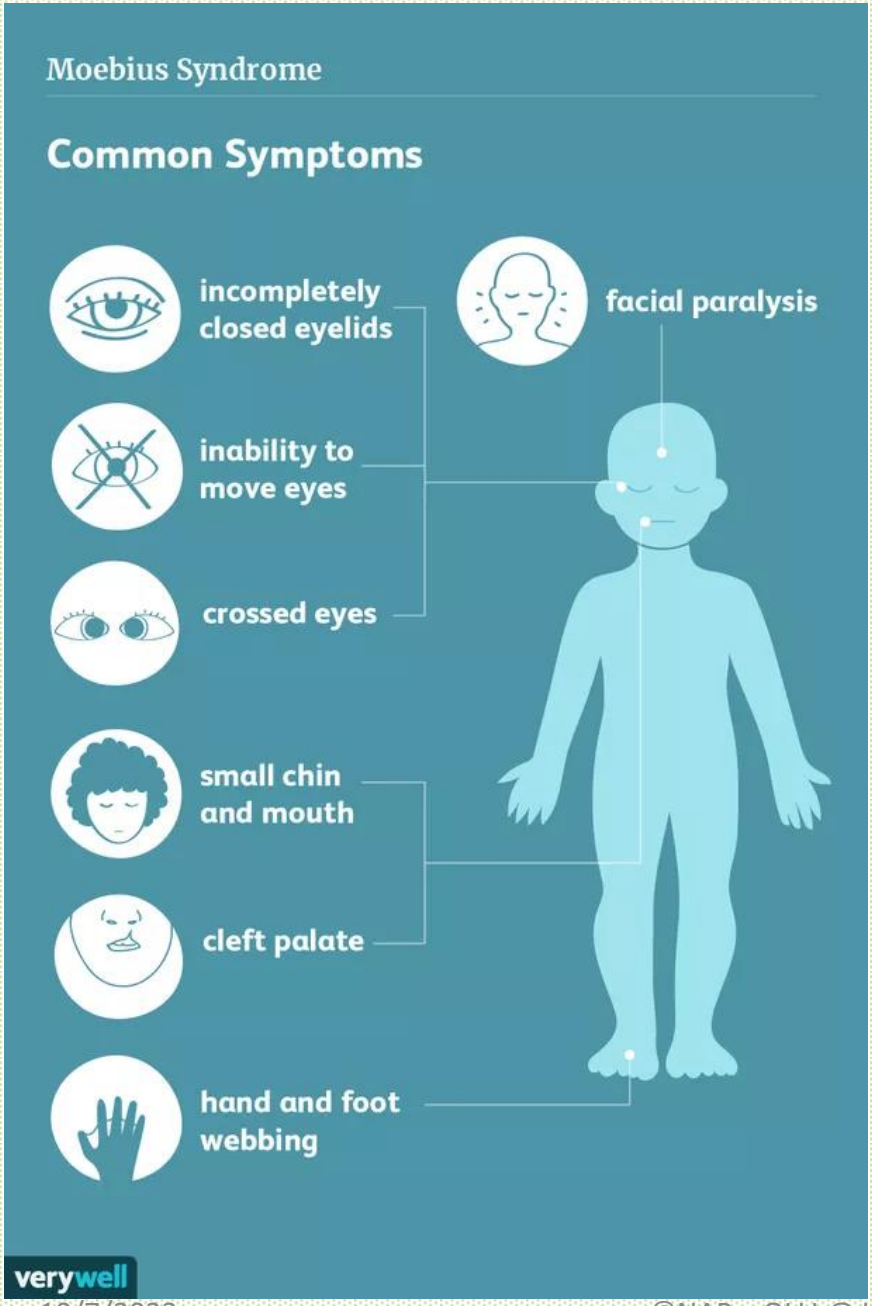
Moebius Syndrome

Other findings

- *Micrognathia* (small chin).
- *Microstomia* (small mouth.
- Short or unusually shaped tongue.
- High, arched or cleft palate.
- Hearing loss.

- Dental abnormalities.
- Strabismus.
- Difficulty making eye contact.
- Bone abnormalities in hands and feet.
- *Hypotonia* (weak tone of muscles).

Moebius Syndrome



Moebius Syndrome

- Delayed development of crawling and walking.
- More likely to have characteristics of autism spectrum disorders ASD (???), but that can be difficult to diagnose in these patients.
- May also be associated with a slight increased risk of intellectual disability, but most affected individuals have normal intelligence.

Moebius Syndrome

- Possibly may result from changes in blood flow to the brainstem during early embryonic development.
- Medications taken during pregnancy and drug abuse (cocaine) may be additional risk factors.
- Less is known about causes of hand and foot abnormalities.

Moebius Syndrome

- Exact incidence is unknown, but it happens in about 1 in 275,000 newborns.
- Causes are unknown, but it probably results from both genetic and environmental factors.
- May be associated with changes in specific regions of chromosomes 3, 10, or 13 in some families.
- A few cases have been reported in families, but most are sporadic and do not show a clear pattern of inheritance.

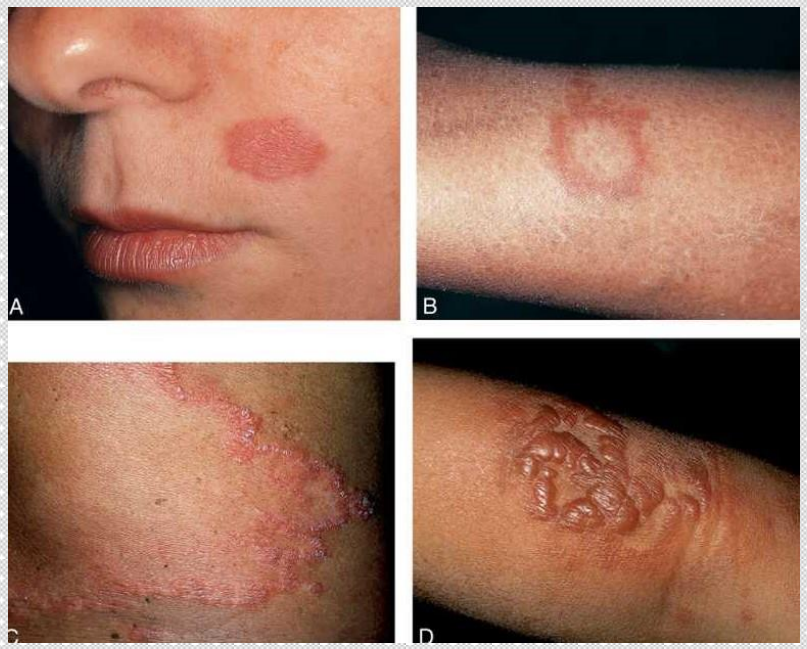
Moebius Syndrome

- Can also affect other cranial nerves that are important for speech, chewing, and swallowing.
- In turn, this affects those functions, which can make quality of life difficult.
- Treatment is possible, but there could be lifelong physical and psychological impairments.

Questions? 2

HANSEN'S DISEASE
(LEPROSY)

Hansen's disease
(Leprosy)



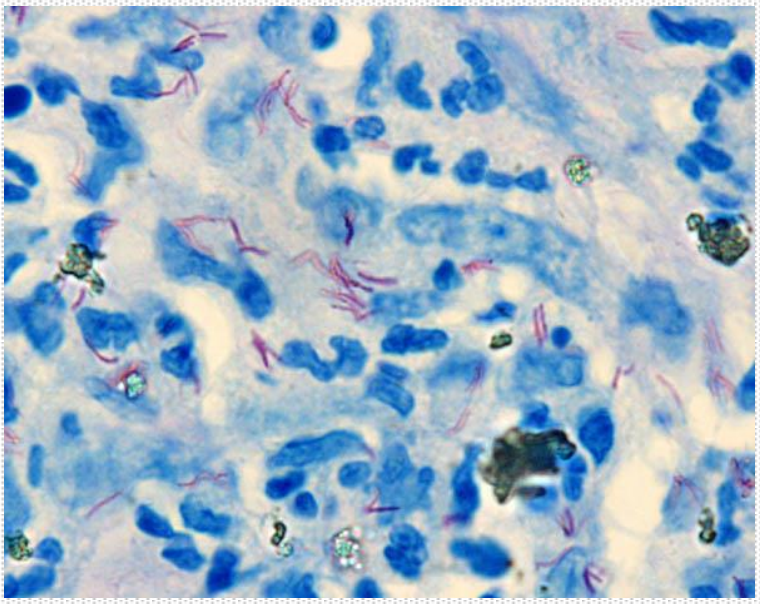
- From the Greek *lepra*, "scale".
- 1st noticeable sign is pale or pink patches of skin that may be insensitive to temperature or pain.
- Sometimes accompanied or preceded by numbness or tenderness in the hands or feet.
- Secondary infections cause tissue loss, deforming and shortening fingers and toes as cartilage is absorbed into the body.

Hansen's disease
(Leprosy)

- Mycobacteriae family:
  - *M. tuberculosis*
  - *M. leprae*
  - *M. lepromatosis (2008)*

- Aerobic acid-fast bacteria, cannot Gram stain.

- *myco* (fungus) refers to the way mycobacteria have been observed to grow in a mold-like fashion on culture surfaces.



Wade-Fite stain

Hansen's disease
(Leprosy)

- M. leprae is an intracellular, aerobic, rod-shaped acid-fast bacterium.

- The leprosy bacteria lack the genes that are necessary for independent growth.

- They are obligate intracellular pathogens, and can not be cultured in the laboratory, but they grow in mice and armadillos.

Hansen's disease
(Leprosy)

- Infection with HIV does not appear to increase the risk of developing leprosy.
- Leprosy is not sexually transmitted and is not spread through pregnancy to the unborn child.
- 95% of people who are exposed to M. leprae do not develop leprosy;

Hansen's disease
(Leprosy)

- Mostly affects and damages the nerves of the skin, the extremities, the lining of the nose, the eyes and the upper respiratory tract .

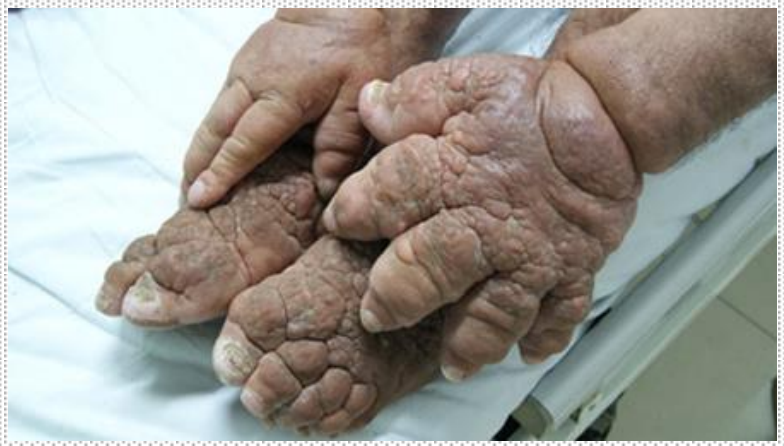
- 3 types:
  - *Tuberculoid (Paucibacillary)*
  - *Lepromatous (Multibacillary)*
  - Dimorphous (Borderline)

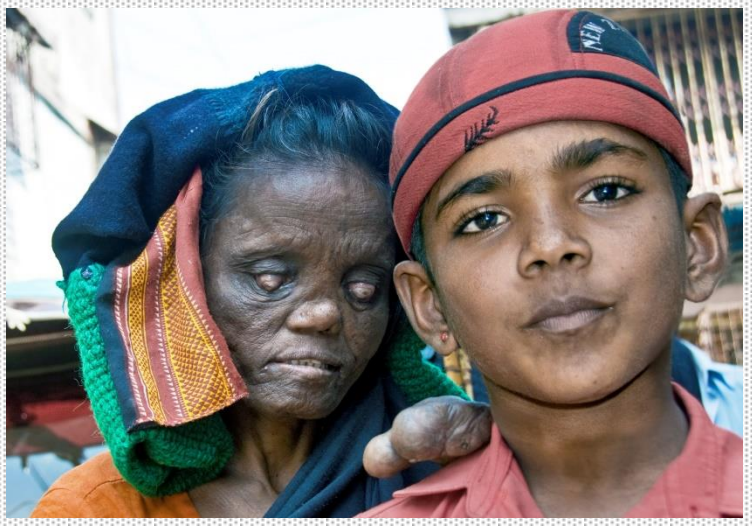
- Treatment stops the disease but reverts nerve injuries only if started early.

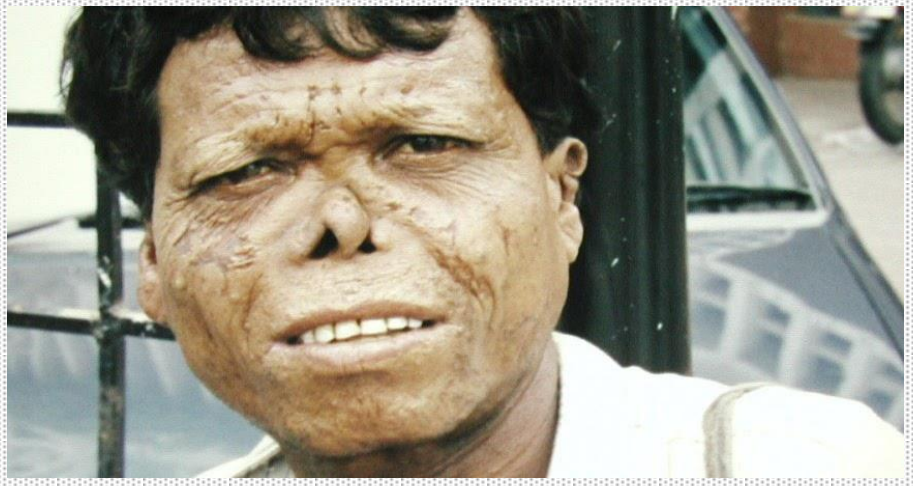
Hansen's disease
(Leprosy)

- The CDC (Centers for Disease Control and Prevention) reports that only 150 to 250 new cases are diagnosed in the US each year.
- Spreads through contact with the mucosal secretions of a person with the infection when the person sneezes or coughs.
- It's not highly contagious, but close, repeated contact with an untreated person for a long period of time can lead to contracting the disease.
- The bacterium multiplies very slowly, and the disease has an average incubation period of five years (WHO), but symptoms may not appear for as long as 20 years.

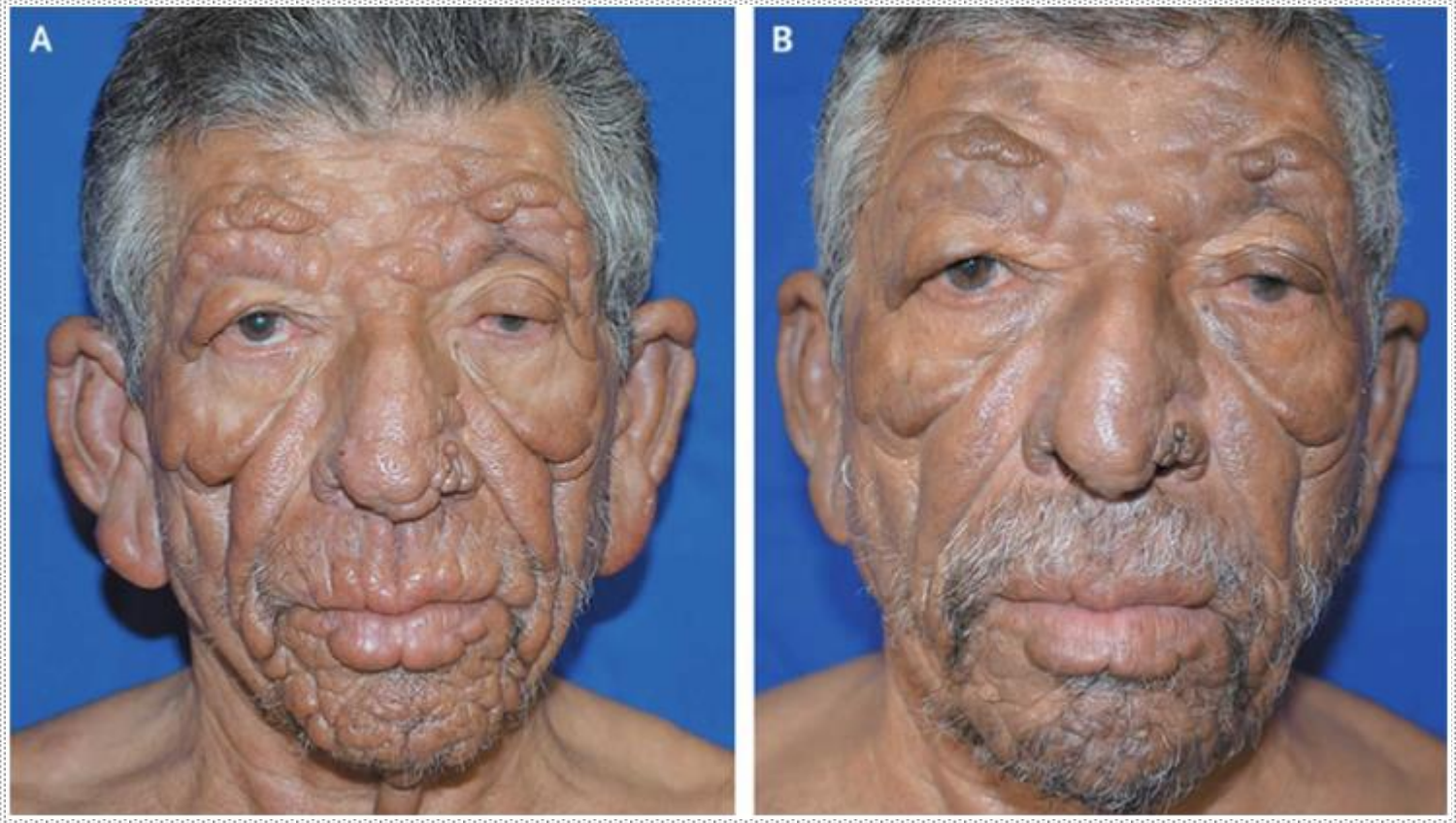
Hansen's disease
(Leprosy)





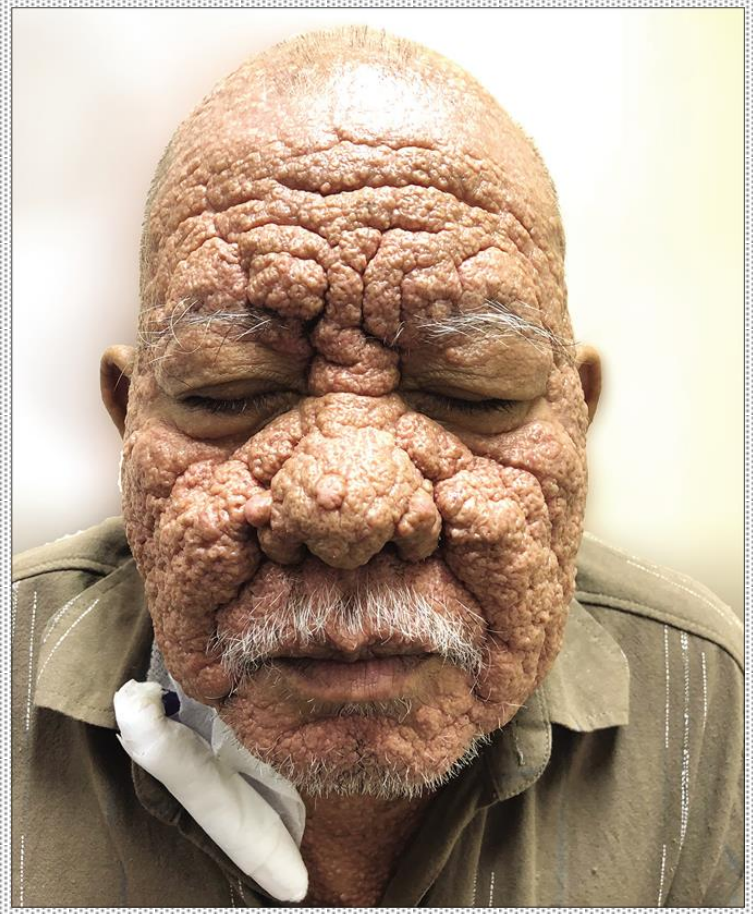


Hansen's disease
(Leprosy)



57 yo man with lepromatous leprosy, before treatment with multidrug (Rifampicin, Clofazimine, and Dapsone) plus prednisone (A), and after 9 months (B). He will need treatment for 24 months, provided free by WHO.

Leonine Facies in Lepromatous Leprosy

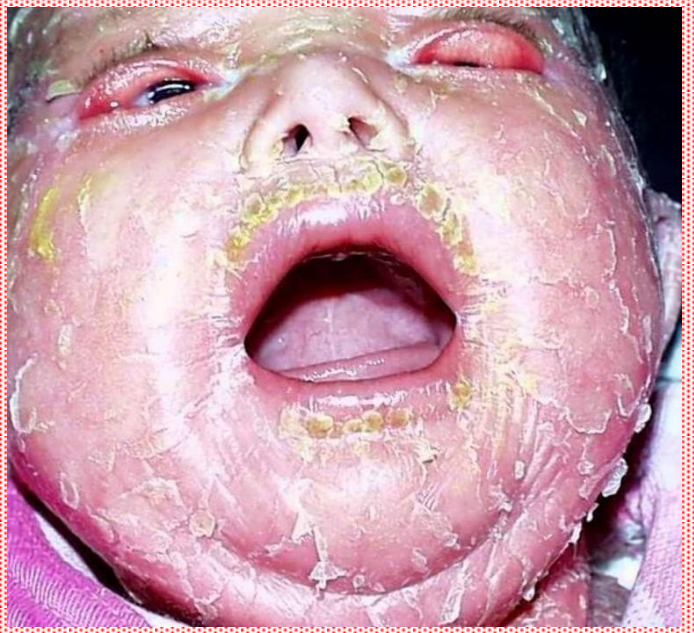


Leonine Facies in Lepromatous Leprosy

ICHTHYOSIS

Ichthyosis

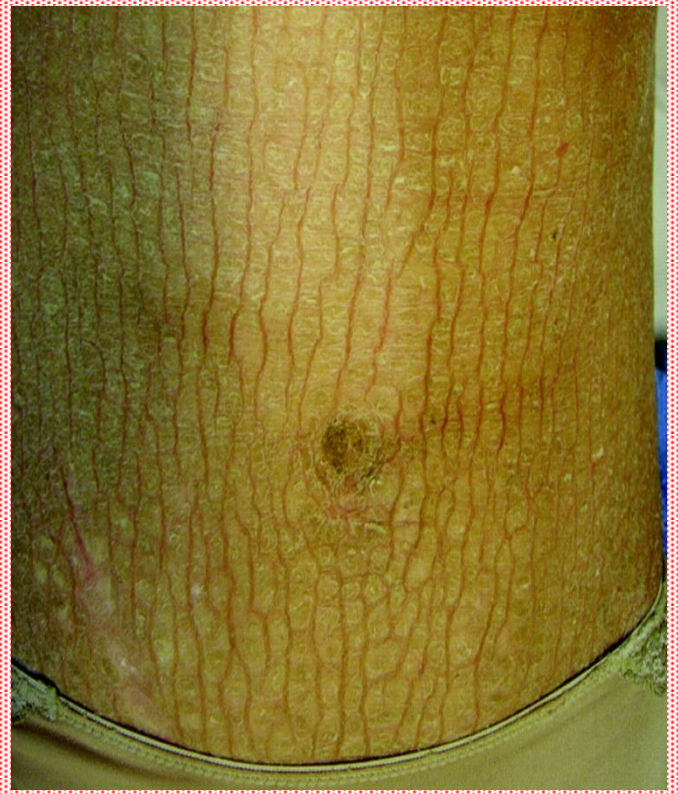
- From the Greek ***ichthys***, "fish“ and ***osis***, “disease” since dry, scaly skin is the feature of all forms of ichthyosis.



- More common in Native American, Asian, and Mongolian groups.
- Autosomal recessive inheritance.
- Mostly skin manifestations but can also have extracutaneous symptoms.

Ichthyosis

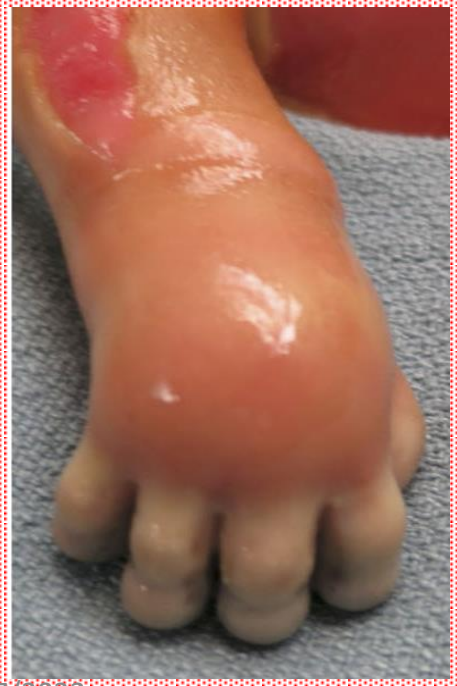
- More than 28 types with a wide range in severity of symptoms and outward appearance.
- Ichthyosis vulgaris accounts for more than 95% of cases
- There can be life-threatening conditions such as harlequin-type (lamellar) ichthyosis.

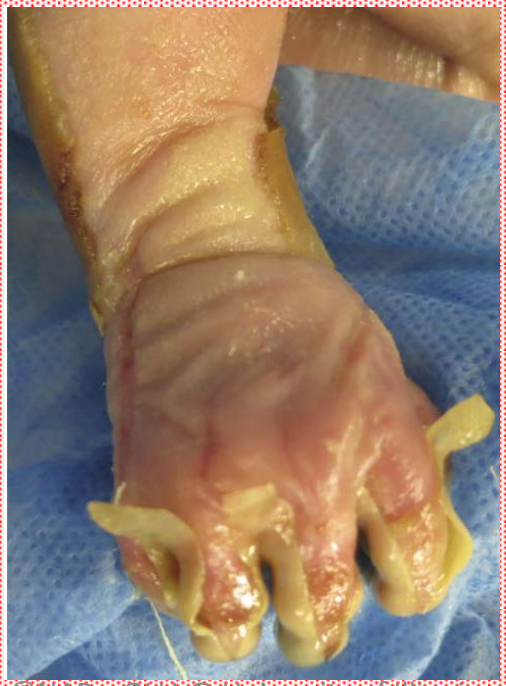


Ichthyosis
(Lamellar)

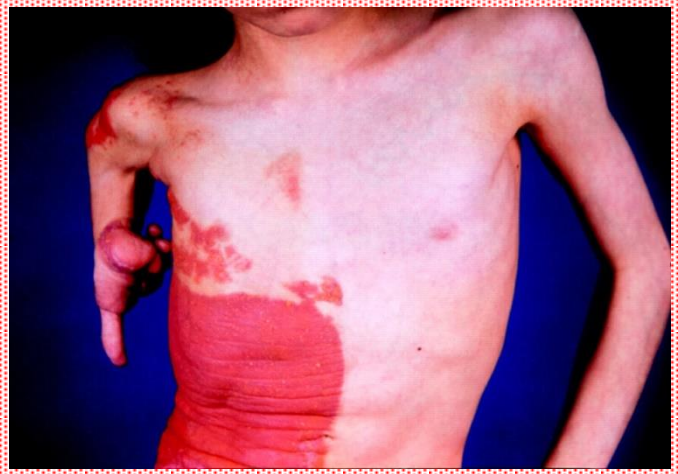
- Sometimes called “collodion baby”.

- Scaly skin is covered by a collodion-like membrane, which dries, scales, cracks and later may bleed.





Ichthyosis



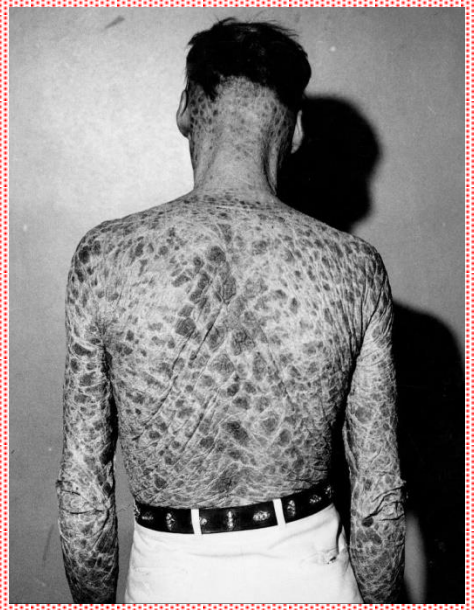
C.H.I.L.D. Syndrome

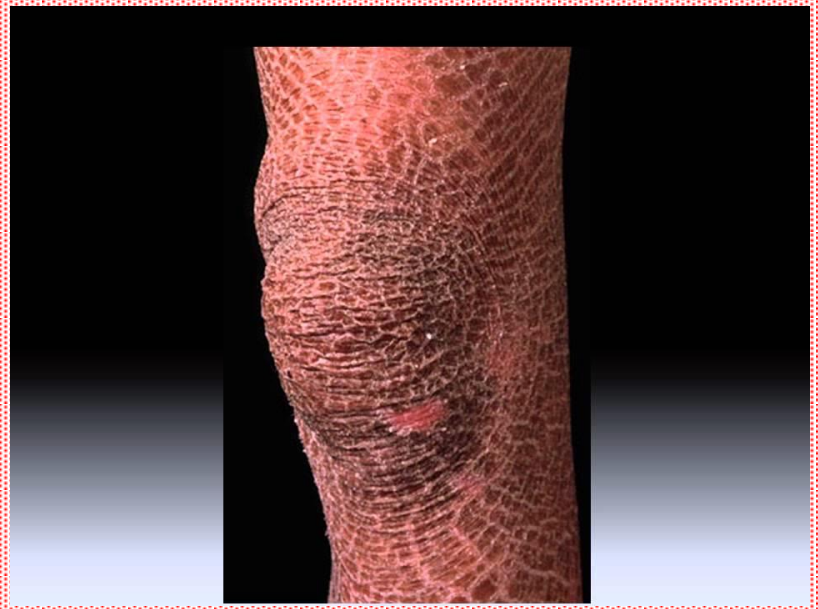
Congenital **H**emidysplasia: mostly in females, the right side of the body is underdeveloped, and internal organs may be affected.

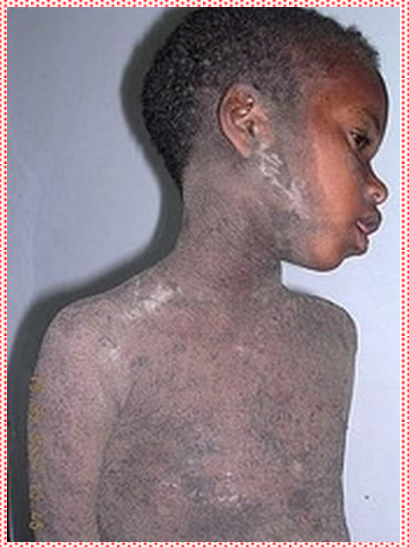
- **I**chthyosiform erythroderma: flaky scales, red patches and possible hair loss on the affected side at birth.
- **L**imb **D**efects—Fingers or toes on the affected side may be missing; an arm or leg may also be shortened or even missing.

Alligator Skin

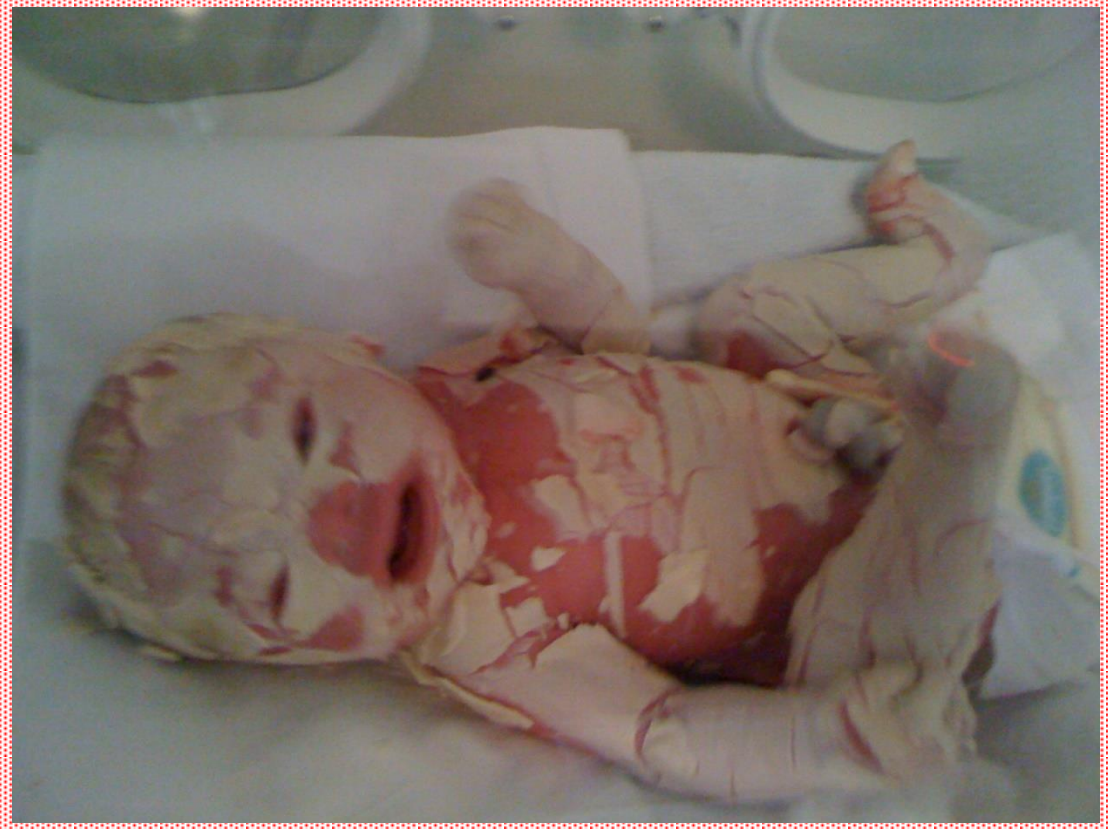
- Hyperkeratotic scaling of the skin with thick plates resembling an alligator hide.
- An autosomal dominant ichthyosis.







Icthyosis
Harlequin Syndrome



CARCINOID SYNDROME

Carcinoid (NET)

When cells grows abnormally, they form a tumor.

Tumors are cancer when they spread within an organ or to other organs.

A NET is a tumor in the neuroendocrine system. *

NET's grow slowly over years, causing few symptoms until they become large or spread to other parts of the body.

NET's can then release hormones like serotonin.

Carcinoid
Symptoms

Skin flushing: Face and upper chest feel hot and change color, from pink to purple; may last a few minutes or a few hours, and may be triggered by stress, exercise, alcohol or unknown causes.

Facial skin lesions: Purplish areas of spiderlike veins appear on nose and upper lip.

Diarrhea: Frequent, watery stools with abdominal cramps.

Difficulty breathing: Wheezing and shortness of breath, may occur with episodes of skin flushing.

Rapid heartbeat: Carcinoid may cause periods of fast heart rate.

Carcinoid

Carcinoid Syndrome (CS)

- Flushing, or redness of the skin, is a common, episodic symptom of CS in ~10% of patients.
- Serotonin and other chemicals released by the tumor cause the skin on the face or neck to feel hot and change color.
- In addition to CS, flushing can be caused by menopause, panic attacks, and alcohol.

Carcinoid

Carcinoid flushing

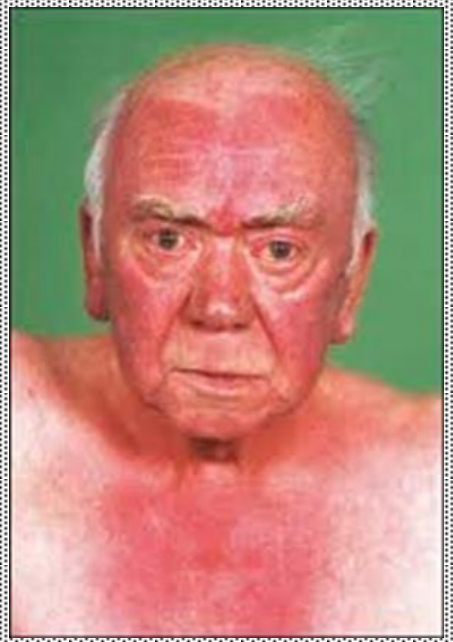
First Type

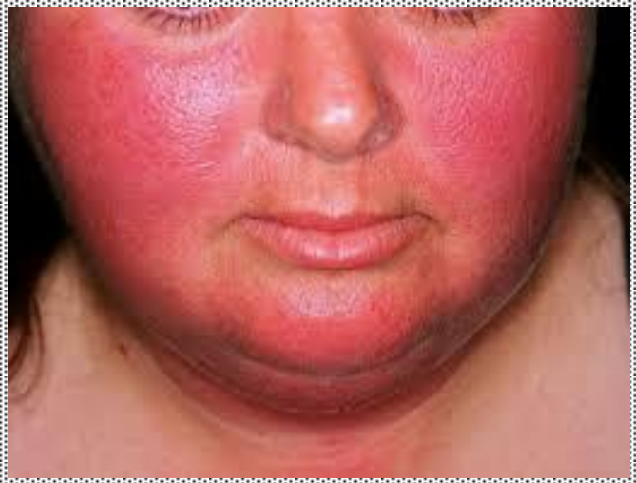
- Faint pink to red, can appear on the face and upper body, and may extend as far as the nipple.
- Initially, it is triggered by alcohol, exercise and food.
- Usually fades within a few minutes, and may occur several times per day.

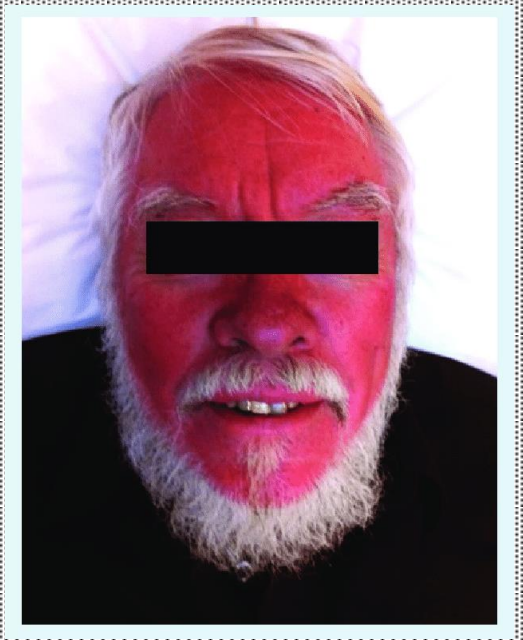
Second Type

- Purple, more intense, often lasts longer and may appear on the upper body and limbs.
- Exact cause is not always obvious, but stress may be a trigger.

Carcinoid Flushing







Carcinoid

Carcinoid Syndrome (CS)

- Serotonin is mainly found in brain, gut, and platelets, it's associated with happiness and optimism (SSRI's), and helps control digestion.
- CS can affect production and storage of serotonin.
- Too much serotonin production can impact a person's physical and emotional well-being.
- After serotonin breaks down in the body, it is normally released in the urine as 5-HIAA.

Carcinoid

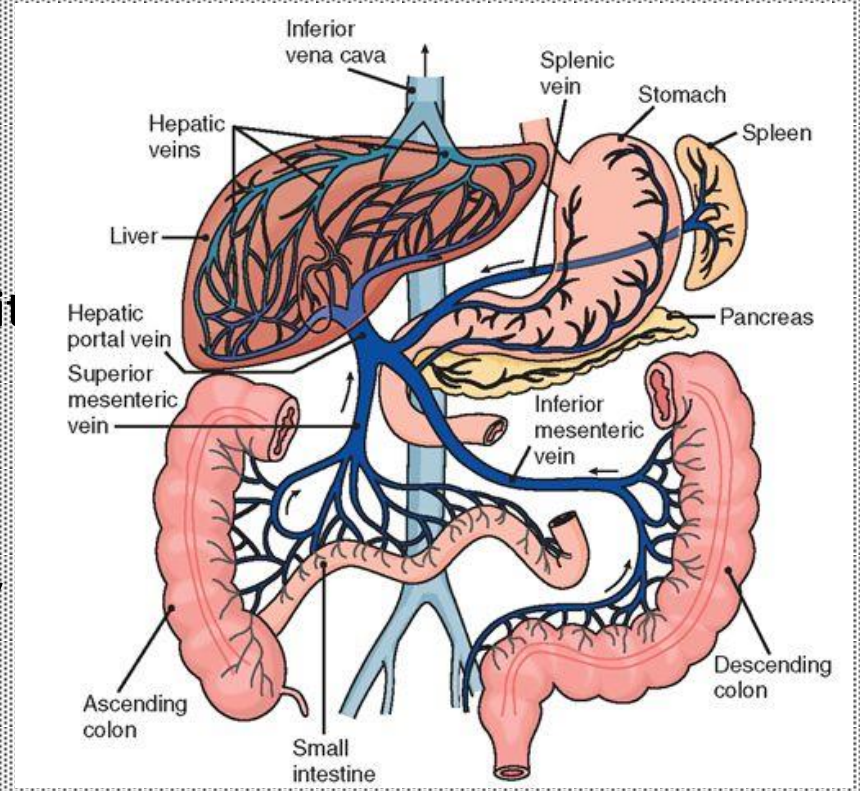
Carcinoid Syndrome (CS)

- Syndrome occurs when rare cancerous tumors (carcinoids), secrete certain chemicals into the circulation, causing various signs and symptoms.
- Neuroendocrine tumors, like carcinoids, occur most often in the GI tract or the lungs.
- To treat CS, treat the cancer, but since most carcinoids don't cause CS until they're advanced, a cure may not be possible.

Carcinoid

Carcinoid Syndrome (CS)

- Some tumors secrete serotonin, but the liver normally neutralizes the chemicals that cause CS before they can cause symptoms.
- **Lung** carcinoid tumors secrete chemicals into the blood upstream from the liver, so it cannot process and eliminate them.
- **Intestinal** carcinoid tumors secrete the chemicals into blood that must first pass through the portal venous system, so they are neutralized before affecting the rest of the body.
- What causes carcinoid tumors is unclear.

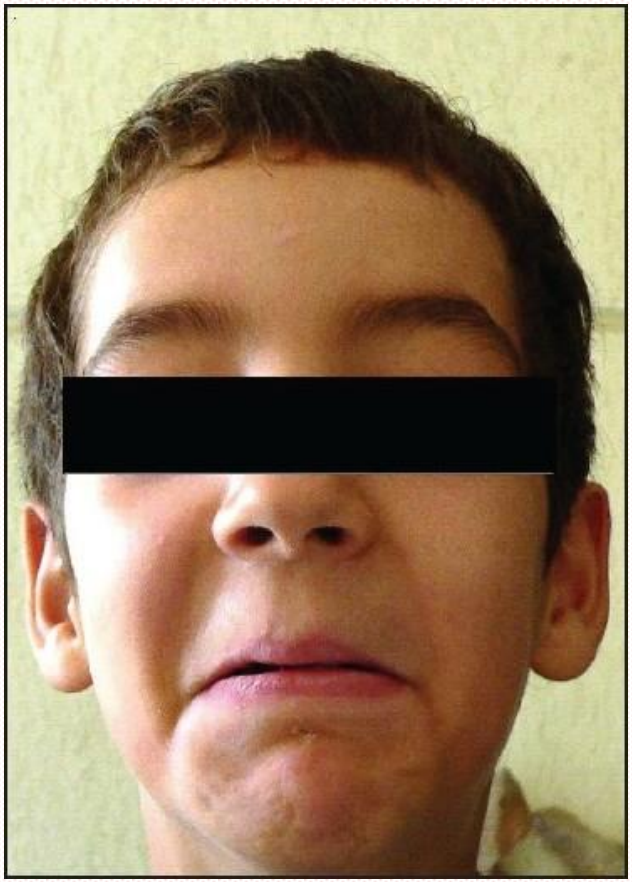


OCHOA SYNDROME
(Urofacial syndrome)

Ochoa Syndrome

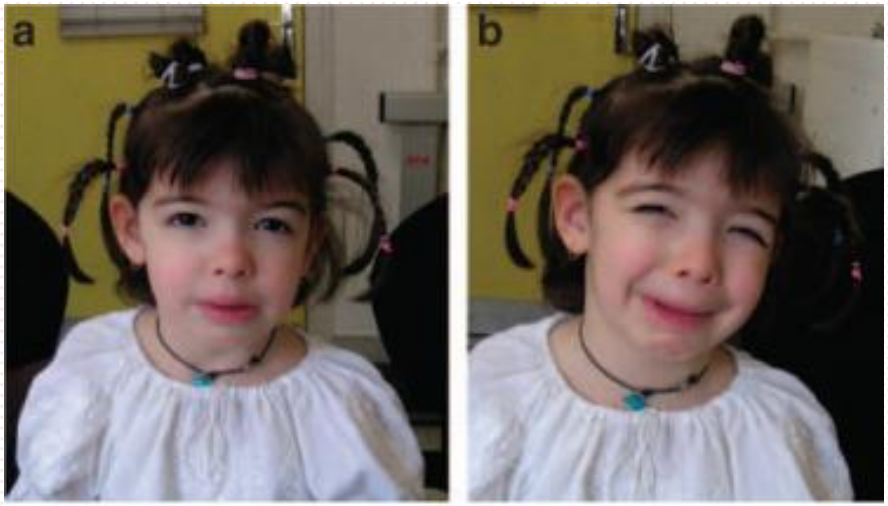
(Urofacial syndrome)

- Autosomal recessive disorder with dysfunction in the lower urinary tract and bowel and with a typical facial expression.
- When attempting to smile, the patient seems to be crying or grimacing.
- First described by the Colombian physician Bernardo Ochoa in the early 1960's.
- Early detection is vital to obtain a better prognosis of urinary symptoms, which can cause serious harm.



Ochoa Syndrome

(Urofacial syndrome)

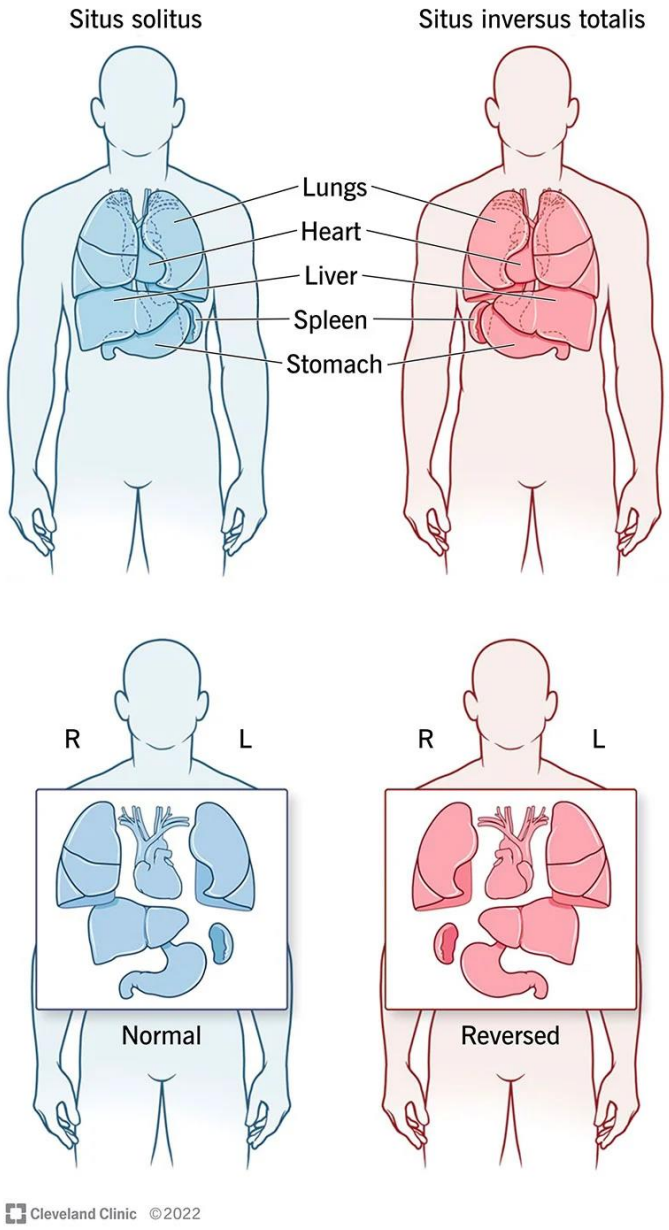


Patient trying to smile

- Symptoms can start at very young age.
- Many patients will die in their teens to early 20's because of renal failure as a result of neurogenic bladder.

- Most present with enuresis, hydronephrosis, urinary tract infection, and X-Ray findings typical of obstructive or neurogenic bladders.

SITUS INVERSUS TOTALIS (SIT)



Situs Inversus Totalis (SIT)

- Organs are arranged on the opposite side of the body.
- Sometimes no side effects.
- Congenital heart defects or other complex issues may be present.
- *SIT* is inherited in an autosomal recessive pattern.
- Frequency of *SIT* is 1/10K and is more frequent in males 1.5 to 1.

Situs Inversus Totalis (SIT)

Heterotaxia (from Greek: *heteros* and *taxis*) is the term for these disorders.

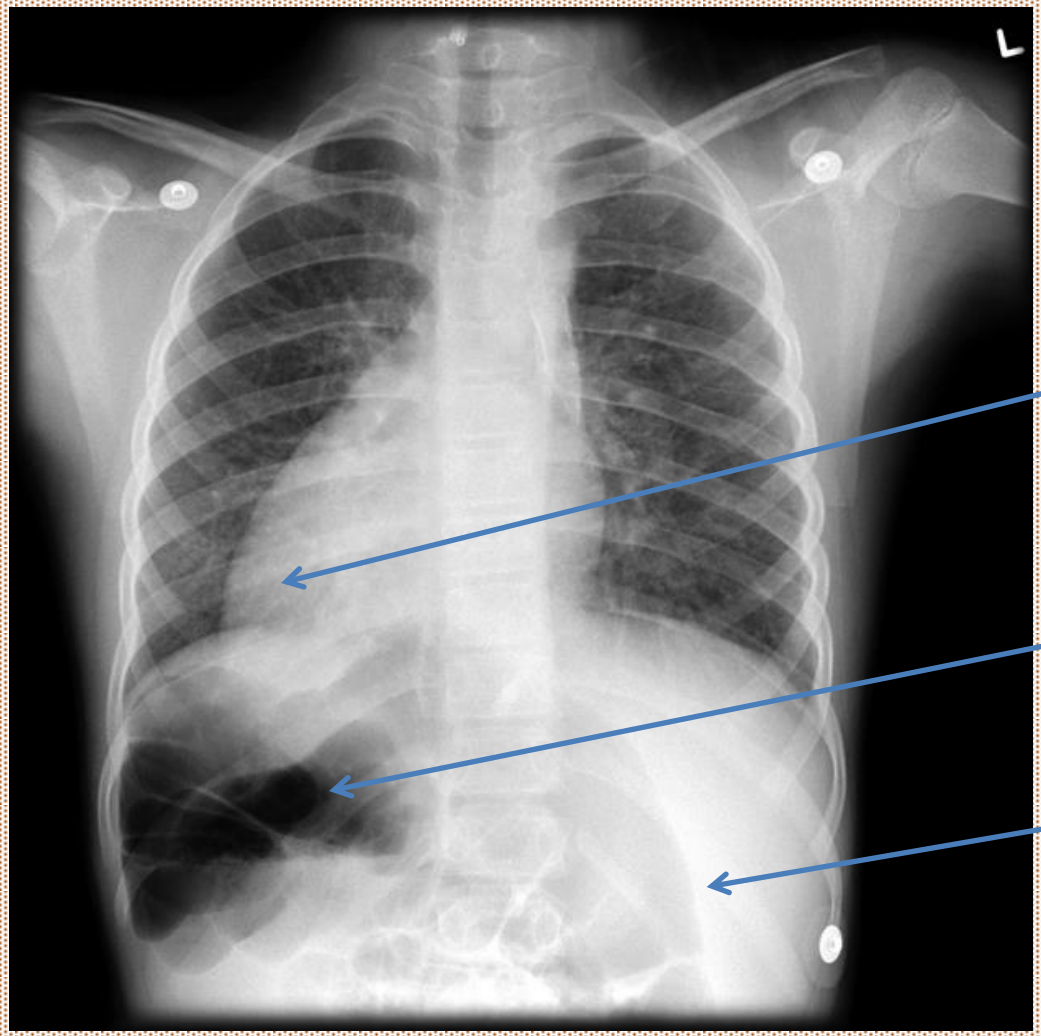
The asymmetry of unpaired organs (heart, liver, spleen, stomach, bowels, aorta, vena cava) is unavoidable.

Paired organs are asymmetrical and have distinct left and right forms.

The lungs have major differences: 3 right lobes vs 2 left lobes.

Vascular differences between L and R kidneys are important for surgeons.

Situs Inversus Totalis (SIT)



Tip of heart is in the Right side.

Stomach air bubble is on the Right.

Liver shadow is on the Left.

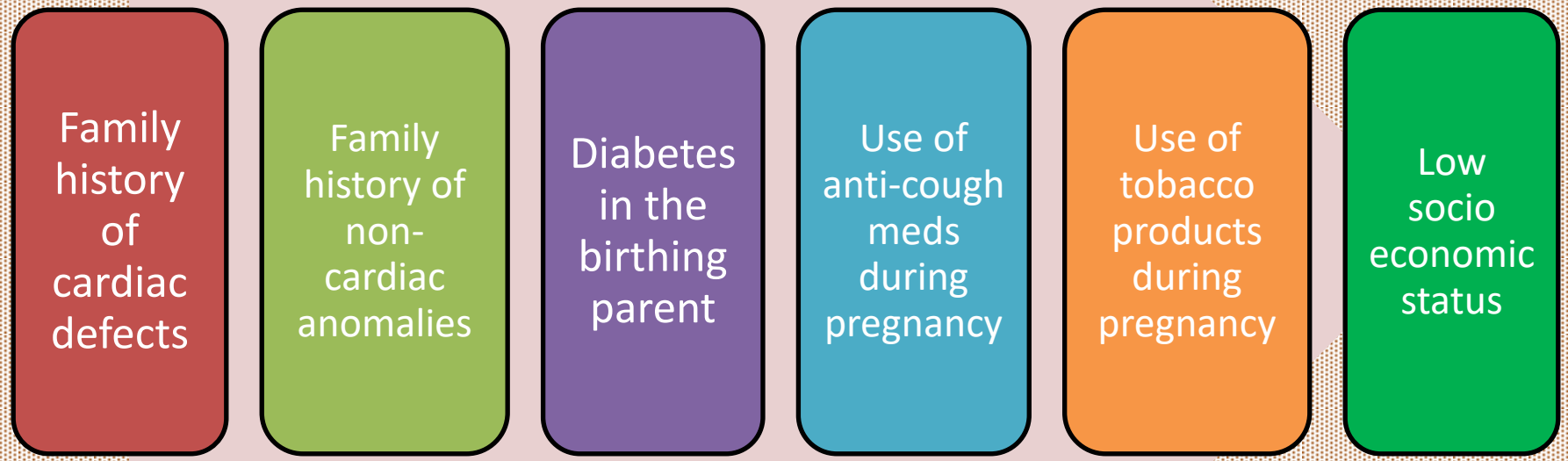
Situs Inversus Totalis (SIT)

- *Normal anatomy is called Situs solitus* (SS).

- *Situs inversus* has two main subtypes:
  - With *dextrocardia*: the point of the heart is on the right side, with the other organs flipped; can be isolated or with cardiac or GI anomalies.
  - With *levocardia*: the point of the heart is on the left side, but the other organs are flipped; more rare, often associated with other cardiac anomalies.

- Another subtype is *situs ambiguous* (SA): several organs are in abnormal positions, but without a defined pattern.

Risk factors for SIT



Situs Inversus Totalis (SIT)

Cough suppressants in Pregnancy

Phenylephrine: not advisable till the end of the first trimester.

Codeine: is not safe in early pregnancy, associated with spina bifida and heart defects in the baby.

NSAIDs: can reduce the amniotic fluid, affecting the growth of the lungs, kidneys, and the baby's heart.

Corticosteroids: not advisable during pregnancy because it may lead to congenital anomalies.

Ethanol: alcohol during pregnancy can have negative consequences such as miscarriage and fetal alcohol spectrum disorders

Sweetening: using a lot of artificial sweeteners while pregnant can cause sugar and blood pressure complications.

Situs Inversus Totalis (SIT)

- Surgeries can be challenging due to "mirror effect":
  - Percutaneous biliary procedures
  - Portal vein embolization
  - Organ transplantation, especially liver & kidneys
  - Cardiac procedures

- Surgical patients with SIT may require more flexibility and creativity from the surgical team.

- Most surgeons are right-handed, so using instruments with the left hand or the pedals with the left foot can be an uncomfortable surgical challenge.

Situs Inversus Totalis (SIT)

- Tell Your Doctor if you have SIT!
- Future ailments can become more difficult to diagnose when you have a mirrored anatomy.
- Appendicitis in normal anatomy presents with pain on the lower right side.
- Your provider may rule out appendicitis if you have pain on your lower left side, which could delay urgent treatment.

Situs Inversus Totalis (SIT)

- The condition is also seen in “mirror image” twins.
- This type of twins occurs when a fertilized embryo splits later than usual during pregnancy.
- Mirror image twins can also be opposites in things like handedness, asymmetrical features, and brain-hemisphere dominance.
- Surgery to reverse the organs’ positioning is usually not recommended.

Final Questions?

MakeAGIF.com