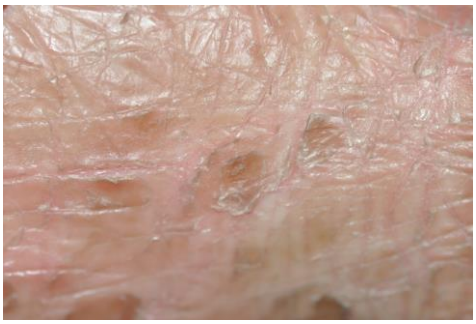
Q 1: What is Ichthyosis?

Ichthyosis is characterized by dry, scaly and thickened skin. (Refer Fig 1.1 & 1.2) It comes from the Greek word meaning 'fish' as the thickened scaly skin sometimes has the appearance of fish scales.



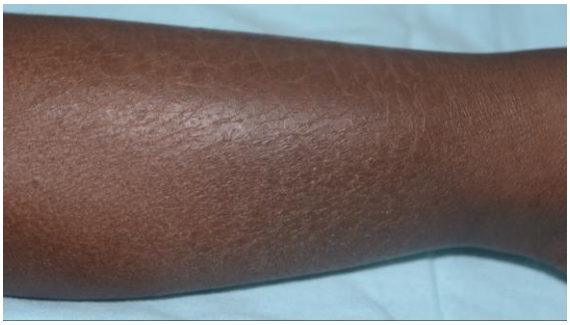


Fig 1 & Fig 2: Dry, scaly and thickened skin

The skin is made up of billions of tiny cells joined together to form our protective covering. It is constantly shedding and being regenerate, and these processes are controlled by genes & modified by environmental factors.

In ichthyosis, it is due to imbalance in the formation of new skin cells & exfoliation of the dead old skin cells. Either way, the end result is ichthyosis. (Refer Fig1.3)

A. High rate of new skin cells formation leading to skin cells are formed at a faster rate than they are needed and they pile up on the skin surface.
B. Low rate of skin cell shedding due to excessively sticky dead skin cells. Skin cells are produced at the normal rate but unable to detach from the cells beneath them when they reach the surface. So they build up in layers.

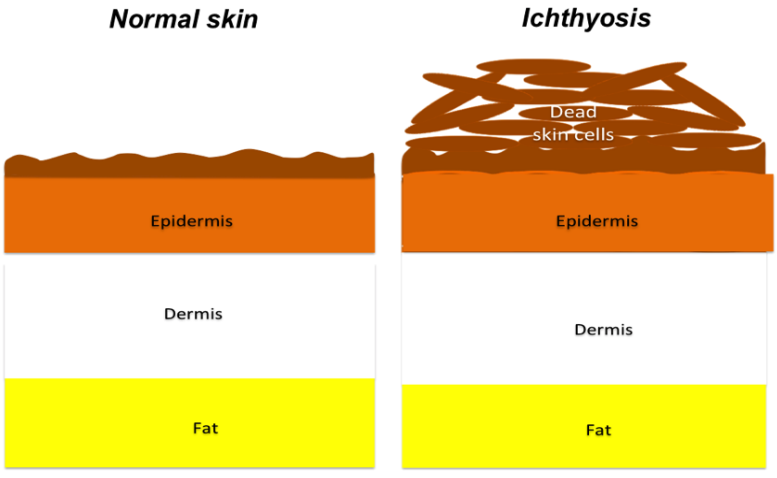


Fig 1.3: Imbalance in the formation of new skin cells & exfoliation of the dead old skin cells

Q 2: What is inherited ichthyosis?

There are at least 20 varieties of ichthyosis, including inherited and acquired forms.

Acquired ichthyosis may occur in later life as the result of medical problems like Hypothyroidism, lymphoma, HIV infection, leprosy and sarcoidosis. Ichthyosis may also be provoked by certain medications: Nicotinic acid, clofazimine & diuretic therapy, etc.

Inherited ichthyosis is usually apparent during the first year of life and continues to affect a person throughout life. Typically, they are inherited from their parents, but sometimes can occur spontaneously in a fetus.

Inherited Ichthyosis is classified based on its severity

A. Mild to Moderate forms (Refer Fig 2.1)

For examples, Ichthyosis Vulgaris, X linked recessive Ichthyosis

The dry scaly skin is usually limited over their extremities & trunk but sparing their faces and flexures. It usually improves with age, regular moisturization and during hot humid weather.

B. Severe forms (Refer Fig 2.2)

For examples, Lamellar Ichthyosis & Non bullous ichthyosiform erythroderma

The dry scaly skin is generalized and usually present at birth.

Fig 2.1: Mild-moderate Ichthyosis

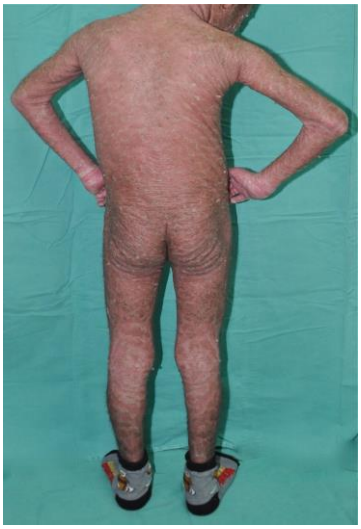


Fig 2.2: Severe Ichthyosis

Q3: What is the cause of inherited Ichthyosis?

Ichthyosis is caused by errors in the DNA code (mutations) that make up genes. These genes are responsible for making and expressing proteins.

Inherited Ichthyosis can result from one genetic mutation in any one of the 13 genes that have been found to cause the isolated hereditary ichthyosis .e.g. Filaggrin gene, tranglutaminase gene, etc. These mutations, or errors in the genetic code, may not allow the affected gene to produce a specific protein that controls the rate of skin cells production & shedding. These mutations in genes passed from one or both parents to a child.

In some cases, (autosomal recessive or X linked recessive ichthyosis) the parents themselves do not have the condition, but carry the mutated genes. In other cases, the mutations may occur spontaneously during the formation of the ovum or sperm by chance.

Q4: Is ichthyosis contagious?

Ichthyosis is not contagious.

It is not caused by a bacteria, virus, or germs. It is not cancer.

People affected by Inherited Ichthyosis are from all races and cultures and in either sex. It is perfectly safe to hug and kiss a person with ichthyosis. You will not get it because Inherited Ichthyosis is an inherited disorder.

Q5: How common is inherited Ichthyosis?

Ichthyosis vulgaris is the commonest form of inherited ichthyosis. It appears in approximately one person in every 250 – 1000 population and often goes undiagnosed because it can be relatively mild.

The severe form of inherited Ichthyosis (Lamellar ichthyosis) is rare. It affects about 1 in 300,000 population. It is estimated that there are about 100 Malaysians live with lamellar Ichthyosis.

Q6: What are the signs and symptoms?

Inherited Ichthyosis is present at birth, but in the milder form (Ichthyosis Vulgaris), symptoms do not become apparent until later in life. The most common symptoms & signs include dry scaly skin, itchiness, excessive skin shedding, overheating, and pain due to loss of skin elasticity and embarrassment. The more severe form of Inherited Ichthyosis (Lamellar Ichthyosis) may present as collodion baby at birth.

Patients suspected of having congenital ichthyosis can consult a qualified dermatologist for proper diagnosis and treatment. The names and place of practice of qualified dermatologists in Malaysia can be found on the Malaysian Association of Dermatologists (Persatuan Dermatology Malaysia) website – www.dermatology.org.my

Q7: What is collodion baby?

The term ‘collodion baby’ is the name given to a baby who is born encased in a skin that resembles a tight and shiny film or sausage skin. (Refer Figure 7.1 & 7.2)

The skin is tight, and this may make the eyelids and lips look as if they are being forced open. The collodion membrane undergoes desquamation, which is usually complete by 2 to 3 weeks of life. This reveals the underlying skin disorder.

Harlequin baby is a severe genetic skin disease, which causes thickening of the outer layer of epidermis (stratum corneum). At birth, the child’s whole body is encased in an 'armour' of thick white plates of skin, separated with deep cracks. In addition, the eyes, ears, penis, and the appendages may be abnormally contracted. (Refer Fig 7.3)

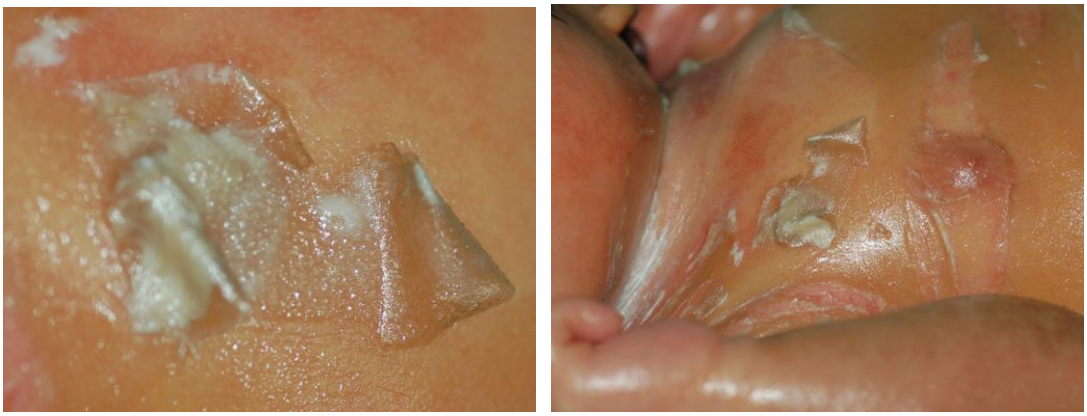


Fig 7.1 & 7.2: Collodion baby

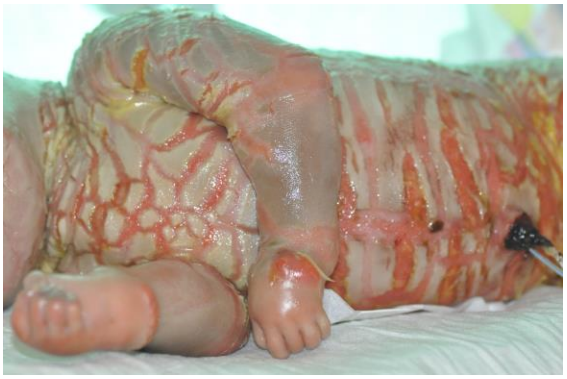


Fig 7.3: Harlequin baby

Q8: Do they have normal lifespan and intelligence?

Most people with inherited ichthyosis have normal lifespan & intelligence.

But they may get complicated by

1. Overheating: ichthyosis may affect normal temperature control by reducing the ability to sweat.
2. Limited movement due to loss of skin elasticity: dry, scaly skin may make it too painful to move some parts of the body.
3. Secondary bacterial infection due to skin cracks.
4. Impaired eyesight due to unable to close their eyes.
5. Social embarrassment: patients with ichthyosis may be subjected to psychological issues including feelings of isolation, low self-esteem, and depression.

Q9: Is there a cure?

There is no cure for the inherited form of ichthyosis at present, but it is possible to manage the symptoms & improve the quality of life of the patients.

The goal in caring for ichthyosis is to hydrate (moisturize) the skin, to reduce scaliness so that the individual is physically and socially comfortable. Vitamin D supplementation is advised for patients with severe forms of ichthyosis.

The following recommendations may be useful:

1. Gentle cleansers (harsh soap may exacerbate dryness)
2. Bathing in baking soda water (one table spoon in 10 liters of water)
3. Rubbing with a pumice stone or exfoliating sponge to remove scale
4. Moisturizing creams with keratolytic feature (urea, salicylic acid, etc)
5. In severe disease, the oral retinoids (Acitretin or Isotretinoin) are helpful

Q10: Are there psychological symptoms associated with ichthyosis?

People with inherited ichthyosis can lead a normal & productive life. However, patients with ichthyosis may be subjected to psychological issues including feelings of isolation, low self-esteem, and depression.

Two of the most difficult times are when a child starts school, and potentially has to deal with the teasing on their own for the first time, and during the teenage years when it is so important not to be different from your peers.

As an adult, he may also experience difficult times when starting college, university, a new job or starting new relationships.

Please contact **PERKIM (Pertubuhan Kebajikan Pesakit Ichthyosis Malaysia; https://www.facebook.com/pertubuhankebajikanpesakitichthyosismalaysia/)** for more information and social supports.

Q11: Is genetic testing available for inherited Ichthyosis?

Yes. Genetic tests are available in Pediatric dermatology unit, Kuala Lumpur General Hospital.

Ichthyosis is usually a clinical diagnosis. But genetic tests can help us to further distinguish the specific types of inherited ichthyosis and the risk in future pregnancy.

Prepared by,

Dr Leong Kin Fon

Consultant Paediatric Dermatologist
Institute of Paediatric, Hospital Kuala Lumpur
Member of Dermatological Society of Malaysia

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