



Epithelial Delicacy Syndrome

Author: Yasir Ali¹; Nida Nawaz²

Affiliation: Minhaj University Lahore¹; University of Sargodha²

E-mail: Yasir.ali6624080@gmail.com¹; baba08050213@gmail.com²

Abstract:

Epithelial delicacy syndrome is an autosomal genetic disease that has swear effects on human body. It raises water filled blisters on hand, mouth, foot and every other skin portion that expose in sun light. In this disease injury on epidermis can't be relief after wash with cold water. It carries gene on X-chromosome so the rate of effect shown equally in male and female.

Keywords: Epithelial delicacy syndrome, epidermis relief, X-chromosome.

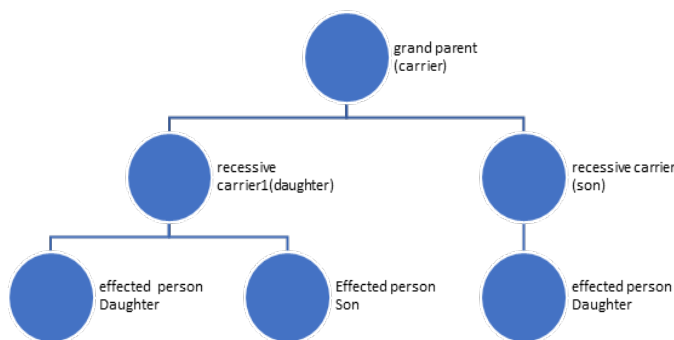
INTRODUCTION

Epithelial delicacy syndrome is an autosomal recessive disease in human from 19th century. The requirement of these observations helps to guide in cure the resulted disease and aware people about its symptoms. Some diseases are genetics and some are autosomal. The cures are resulted to control of wide spread them and save human life. The sun radiations are damage human and animal bodies in different points and body parts .skin diseases are kindler syndrome [1], sun burn poikiloderma and

tissue damage. The related syndrome is under observation and experimentation to find medicine to control them and control to prevent more damage. Here are discussed the epithelial [2] delicacy after facing the sun

exposure. Skin is damage after sun rays IR to strike skin outer layer. Rays destroy genes of chromosomes²⁰ of short arm of skin protective melanin [3]. Hairs growth increase and skin is delicate even a touch can cause a scare and cause of water filled bubbles on epidermis layer. Skin damage effect the whole activities of life like touch, hold, capture etc. the resultant observations help to indicate disease. Different dermatologist gives it different name different medicines are prescribes to control its further spreading. kindler syndrome was first described by Theresa Kindler in 1954 6-year-old girl. It is a rare autosomal recessive Geno dermatosis characterized as dermal syndrome. In it is difficult to understand which aspect of integrin function is at fault in skin. Clinical observations are only to line up the data of different effected families in world. A large number of effected persons are reported in

Pakistan second case in Pakistan (Sialkot) in 10 year old girl in 26 October 2006.the number of person of one effected family are 3 which are recessive genotypic from parents to their offspring.



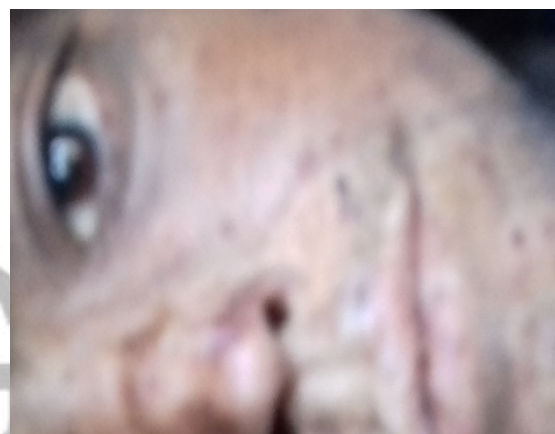
There is a hierarchical data of one of effected and recessive carrier in Pakistan facing the problem of epithelial delicacy syndrome. according to this cross linkage the female recessive carrier causes more effect in dominant person with epithelial syndrome. The male carrier cause of a smaller number of dominant cases



figure; clinical result of effected hands palm with blisters after sun exposure in kindler syndrome. Damage cause the pain in hands with blisters and epidermal layer removing effects. A 30 plus year man.



figure; Effected hand with uv radiations of sun and shown epithelial delicacy hydromania removed, blistering and permanent scars after rupture of blisters or cutoff.



figure; A girl that has symptoms and strongly face the disastrous effect of hyper melanin and basic dermal layer fluid toward ectoderm and blistering with sun exposure.

Epithelial delicacy is not understood clearly because of the related effects and results with kindler syndrome and poikilodermas around the world. It is significant to raise the awareness in people to control the rate of dermal effects in new generation. it must be clinically consulted to control resulted expressions after blistering start the more the exposure to sun light the more damaged the skin and immune lost. Many physicians have no idea about that syndrome they only recommend any other ointments to take on blisters and scars. There is no report is conducted to indicate the exact results of epithelial delicacy syndrome. In past time, a

case of epithelial delicacy syndrome was reported but it confused with other skin diseases like kindler syndrome. The intensity of disease is less some time but it doesn't completely cure and stop to spread.

Symptoms

Itching in hands and face after exposure to sun. Redness on skin after scratching on hands and any area of body. Swelling in eyes. On birth time eyes filled with yellow pus. Time to show symptoms is vary in some patients it starts in 3 three-year-old and in some after 5 years old age. Thinning of skin known as atrophy. A major reason of this syndrome. Swear pain in skin point of effected body part. Swelling of lymph node in arm pit and leg pit. Muscle weakness.

Effects

It causes the eyes burn and cause problems in sunlight sight. Mild skin layer that even a simple pinch removes epidermal layer and produce a uncontrolled growth blister which increase in size after few minutes. Urine turn into red or yellow colored excretory fluid. Permanent scars are raised on effected area of body (firstly white then black) Antibiotics use by unaware person cause teeth and bones yellow.

Un control growth of hairs on mouth and arms including other effected areas of body. It also cause of lack of social activities due to lack of confidence on damaged mouth and hands. Veins of blood are shrink due to effects of scars and blisters. Stunted growth of hands if more duration give to cure it. Joints are not properly work even not able in movement. Protruded nails and dark brown coloration. It also cause of nail remove from finger tips and feet.

Experiment

The experimental procedure is conduct in a girl who shown strong symptoms related to syndrome. The case study is reported by a doctor who was in practical field of skin diseases in CMH hospital Sialkot (Pakistan). The patient of this disease is a 10 year old female. only observation of physician indicate this disease after some x-ray analysis in laboratory.

When dominant disease carrier faces an exposure to sun in morning the effect of sun radiations cause itching on skin of patient. After some time duration of half an hour the results will show with redness in skin with water filled blisters. it is a painful time when blisters are grown larger in number and size .if don't cut blisters they become larger even spread in whole hand or expose body part. The blisters turn into red colored fluid filled in it if not grow larger in size . the effect of these blisters cause of lymph node swelling of related point of attack of radiations.

If cut these resulted blisters then wound take even a month of time to cure and painful effects. These leave spots of white color which turn int black color. Immunofluorescence and gene expression are used to find locus of effected area and photosensitivity of body. It reveals the rate of epidermolysis on Basel membrane. It gives different stain with delicacy of Epithelial cells. Cell adhesion lose due to which blistering become started.

Result

Epithelial delicacy syndrome is not curable disease. But only way to control by using sunblock and wear gloves and avoid to exposure toward sunlight all time and every season. Some ointment relief to blisters but not permanently cure this. Some

laboratory experiment may helpful to control this syndrome in future.

Conclusion

About more than five decade the skin disease are under examination of many dermatological laboratories and clinical observational centers. Different results shown to effect of ultra-radiation, sunlight. This is highly different from other epidermolysis bolus. It increase mucocutaneous rate in skin.

References

- 1 Kindler T. Congenital poikiloderma with traumatic bulla formation and progressive cutaneous atrophy. *Br J Dermatol* 1954; 66: 104–11.
- 2 Weary PE, Manley WF, Graham GF. Hereditary acrokeratotic poikiloderma. *Arch Derm* 1971; 103: 409–22.
- 3 Forman AB, Prendiville JS, Esterly NB et al. Kindler syndrome: report of two cases and review of the literature. *Pediatr Dermatol* 1989; 6: 91–101.
- 4 Shimizu H, Sato M, Ban M et al. Immunohistochemical, ultrastructural, and molecular features of Kindler syndrome distinguish it from dystrophic epidermolysis bullosa. *Arch Dermatol* 1997; 133: 1111–7.
- 5 Yasukawa K, Sato-Matsumura KC, McMillan J et al. Exclusion of COL7A1 mutation in Kindler syndrome. *J Am Acad Dermatol* 2002; 46: 447–50.
- 6 Jobard F, Bouadjar B, Caux F et al. Identification of mutations in a new gene encoding a FERM family protein with a pleckstrin homology domain in Kindler syndrome. *Hum Mol Genet* 2003; 12: 925–35.
- 7 Siegel DH, Ashton GH, Penagos HG et al. Loss of kindlin-1, a human homolog of the *Caenorhabditis elegans* actinextracellular-matrix linker protein UNC-112, causes Kindler syndrome. *Am J Hum Genet* 2003; 73: 174–87.
- 8 Ashton GHS, McLean WHI, South AP et al. Recurrent mutations in kindlin-1, a novel keratinocyte focal contact protein, in the autosomal recessive skin fragility and photosensitivity disorder, Kindler syndrome. *J Invest Dermatol* 2004 (in press).