Hereditary Poikilodermatous Syndromes

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Several diseases promote aging of the skin, which may present in early childhood or adulthood. Most of these conditions are genetically inherited though some, which are acquired, may produce the same picture by affecting the connective tissue. Often these conditions are characterized by multisystems involvement and may remain unrecognized particularly when seen by those in specialties than dermatology. One important group which can present to the dermatologist with this clinical appearance constitutes poikiloderma. This includes a number of disorders, often inherited. All these conditions present with pigmentary changes, atrophy of the skin and telangiectases. (Fig.1). Hereditary diseases presenting with poikiloderma are relatively rare. Proper diagnosis depends on clinical examination and follow-up. Apart from involvement of other systems, some of these conditions are associated with increased risk of cancers. Treatment is multidisciplinary, often difficult and only palliative.

We intend to give a panoramic view of the poikilodermatous entities laying emphasis on the differentiating features between them.

The main disorders that present with poikiloderma are:

- Rothmund – Thomson Syndrome
- Dyskeratosis Congenita
- Xeroderma Pigmentosum
- Kindler Syndrome
- Epidermolysis Bullosa Simplex Variants

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Table 1 provides a synopsis for the main diseases in this group.

Disease	Inheri-tance	Genetic defects	Time of Onset with Symptoms	Skin	Hairs	Nails	Teeth & Mucous Membranes	Others
Rothmund Thomson syndrome (Rothmund 1868)	A.R.	DNA helicase gene RECQL4 on chromo-some 8q24.3 in some cases	3-6 months Erythema & blisters on face & limbs	Poikiloderma	Sparse	In 25% of patients, dystrophic nails are reported	Abnormal dentition	Cataract and skeletal anomalies
Xeroderma pigmenosum (Jkaposi 1874)	A.R.	DNA repair defects, there are different complementa-tion groups (A-F) with variable symptoms and degrees of severity	6 months- 3 years Erythema & freckling	Neoplasia & photo-sensitivity	-	-	Oral carcinoma	Eye & central nervous system abnormalities
Dyskeratosis congenital (Zinsser 1906)	X-linked recessive A.D.	Dyskerin gene on chromo-some Xp28	5 – 13 years Nail dystrophy	Poikiloderma (face, neck, and trunk)	Normal, but thinning alopecia of the scalp, eyebrows and eyelashes has been reported	Dystrophic with ridges, a trophic or absent	Premalignant leukoplakia of mucosal surfaces. Also, leukokeratoses leading to anorectal & urethral stenosis	Hematologi-cal disorders & neoplasia
Kindler syndrome (hereditary acrokeratotic poikiloderma) (Kindler 1954)	A.R.	KINDI gene on chromo-some 20p13	Blisters at birth	Photosensi-tivity & webbing of fingers	-	+	Periodonti-tis, leukokeratoses, and urethral stenosis	-
Weary syndrome (hereditary acrokeratotic poikilo-derma, weary type) (Weary et al, 1971)	A.D.	Not known	Vesicopustules of hands at infancy	Eczema, Sclerotic bands and punctate palmoplantar keratoderma	-	-	Periodontitis	-

AD: Autosomal dominant
AR: Autosomal recessive
Figure1: Poikiloderma in Kindler syndrome (Fig.1a) mild (trunk) and (Fig.1b) severe (forearm) in 2 different patients.