GENES AND PHENOTYPES INVOLVED IN AUTOSOMAL RECESSIVE ICHTHYOSIS: A CLINICAL AND GENETICAL REVIEW

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Abstract
Ichthyosis is a large heterogeneous group of genetic anomalies of skin which are distinguished by either distribution or quality of scaling and hyperkeratosis, also may contain other extra cutaneous and dermatoLOGIC involvement. These anomalies are due to gene sequence variants often linked in skin barrier formation. Inherited ichthyoses can be of either two forms; non syndromic and syndromic forms. Non syndromic ichthyoses only have phenotypic expression causing effect to skin. Non syndromic ichthyoses consists of autosomal recessive congenital ichthyoses, X-linked ichthyoses, ichthyoses vulgaris. Keratinopathic ichthyoses and other forms. This review emphasis on updates of all types of non-syndromic ichthyoses. This review figures out the molecular pathways and phenotype/ genotype associations. Autosomal recessive congenital ichthyoses can be of either three major phenotypes (lamellar ichthyoses, congenital ichthyosiform erythroderma and harlequin ichthyoses) more over, and be three of the minor subtypes (acral self-healing collodion baby, self-healing collodion baby and bathing suit ichthyoses). Keratinopathic ichthyoses are caused due to keratin genes sequence variant so are termed Keratinopithic ichthyoses. For diagnoses and discovery of ichthyoses genetic causes, the next generation sequencing is considered the influential tool. This paper reviews the recent pathomechanisms for the non-syndromic ichthyoses and explains the future perspectives

Keywords: Autosomal Recessive, Ichthyoses, Skin, Genes

INTRODUCTION
Ichthyosis is an inherited genetic deformation characterized by cornification of skin. It may be inherited as autosomal recessive, X linked and autosomal dominant fashion. Clinically ichthyosis is characterized by scaling of skin, which may sometimes be associated with non-cutaneous and cutaneous malformations including palmoplantar keratoderma, recurrent infections, erythroderma and hypohidrosis (1). Ichthyosis may be caused by more than fifty genes sequence variant, it degrades the intra-cellular lipid biosynthesis pathway, DNA repair, adhesion and desquamation and also many other pathways. Each feature interrupted barrier function, even many pathways included pathogenesis (2). clinical malformations, mode of genetically inheritance of a consensus classification system for ichthyosis have been established based on the pathophysiology which is established by (3). Additionally, based on oji classification, ichthyosis can be classified into two main types; non syndromic and syndromic one, the non-syndromic