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Clinical and Molecular Characterization of Ichthyosis at King Abdulaziz Medical City, Riyadh KSA

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Abstract : Ichthyosis is a disorder of abnormal keratinization, characterized by excessive scaling, and consists of more than twenty subtypes varied in severity, mode of inheritance, and the genes involved. There is insufficient data in the literature about the epidemiology and characteristics of ichthyosis locally. Our aim is to identify the histopathological features and genetic profile of ichthyosis. Method: It is an observational retrospective case series study conducted in March 2020, included all patients who were diagnosed with Ichthyosis and confirmed by histological and molecular findings over the last 20 years in King Abdulaziz Medical City (KAMC), Riyadh, Saudi Arabia. Molecular analysis was performed by testing genomic DNA and checking genetic variations using the AmpliSeq panel. All disease-causing variants were checked against HGMD, ClinVar, Genome Aggregation Database (gnomAD), and Exome Aggregation Consortium (ExAC) databases. Result: A total of 60 cases of Ichthyosis were identified with a mean age of 13 ± 9.2. There is an almost equal distribution between female patients 29 (48%) and males 31 (52%). The majority of them were Saudis, 94%. More than half of patients presented with general scaling 33 (55%), followed by dryness and coarse skin 19 (31.6%) and hyperlinearity 5 (8.33%). Family history and history of consanguinity were seen in 26 (43.3%), 13 (22%), respectively. History of colloidal babies was found in 6 (10%) cases of ichthyosis. The most frequent genes were ALOX12B, ALOXE3, CERS3, CYP4F22, DOLK, FLG2, GJB2, PNPLA1, SLC27A4, SPINK5, STS, SUMF1, TGM1, TGM5, VPS33B. Most frequent variations were detected in CYP4F22 in 16 cases (26.6%) followed by ALOXE3 6 (10%) and STS 6 (10%) then TGM1 5 (8.3) and ALOX12B 5 (8.3). The analysis of molecular genetic identified 23 different genetic variations in the genes of ichthyosis, of which 13 were novel mutations. Homozygous mutations were detected in the majority of ichthyosis cases, 54 (90%), and only 1 case was heterozygous. Few cases, 4 (6.6%) had an unknown type of ichthyosis with a negative genetic result. Conclusion: 13 novel mutations were discovered. Also, about half of ichthyosis patients had a positive history of consanguinity.

Keywords : ichthyosis, genetic profile, molecular characterization, congenital ichthyosis
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