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p63 AND DLX RELATIONSHIP: RELEVANCE IN HUMAN HEREDITARY ECTODERMAL DYSPLASIA SYNDROMES

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p63, hereditary disease, epithelial development and differentiation

DLX and p63 proteins are families of transcription factors that plays a pivotal role in many developmental processes, ranging from organization of the body plan to differentiation of individual tissues. Mutations in the p63 and DLX genes are responsible for Ectodermal Dysplasia Syndromes (EDs), a group of pathological conditions that share common anomalies in epithelial- and mesenchimal-derived organs such as hair, tooth, nails, and sweat glands and have been associated with abnormalities in other organs (Priolo and Lagana, 2001). DLX3 mutations are responsible for tricho-dento osseous syndrome (TDO) and amelogenesis imperfecta, hypoplastic-hypomaturation, with taurodontism (AIHHT), while mutations in the p63 gene have been associated with ectrodactyly-ectodermal dysplasia cleft lip/palate (EEC), ankyloblepharon-ectodermal dysplasia clefting syndrome (AEC), and split hand/foot malformation (SHFM). We and others have shown that p63 acts upstream of the Dlx genes in a transcriptional regulatory pathway relevant for ectodermal dysplasias. More recently, we have found that DLX3 and p63 proteins can interact and exert a reciprocal regulation on their activities. We propose that DLX genes and p63 are components of multiple regulatory mechanisms and signaling pathways that play a crucial role during development and differentiation.