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INTRODUCTION

The *CDH1* gene encodes E-cadherin, a calcium-dependent cell adhesion molecule that is essential for the formation and maintenance of intercellular junctions. E-cadherin plays a critical role in preserving epithelial cell integrity and polarity by mediating cell-cell adhesion. Clinical and genetic evidence suggests that *CDH1* is associated with susceptibility to certain types of cancer as well as orofacial clefts.

OBJETIVE

The objective of this study was to conduct an in-silico analysis of *CDH1* nonsynonymous variants (SNVs) associated with cancer and orofacial clefts.

METODOLOGY

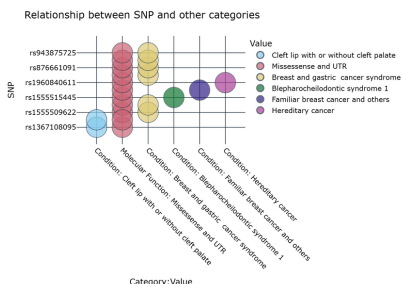
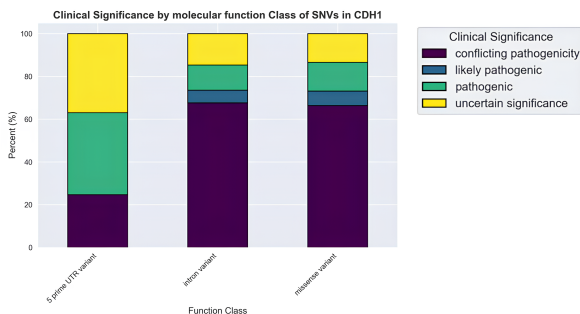
dbSNP

PATHOGENIC VARIANTS IN THE CODING AND REGULATORY REGIONS OF THE *CDH1* GENE WERE INCLUDED; BENIGN AND SYNONYMOUS VARIANTS WERE EXCLUDED.

ClinVar

PATHOGENIC *CDH1* VARIANTS ASSOCIATED WITH DISEASE WERE INCLUDED (MISSENSE AND UTR), WHILE NONSENSE AND BENIGN/SYNONYMOUS VARIANTS WERE EXCLUDED.

RESULTS



DISCUSSION

The SNVs identified in the coding region of the *CDH1* gene suggest a strong functional link between cancer and orofacial clefts. Although these SNVs have not yet been validated through experimental analyses, and their associations are currently based on in silico predictions, several studies have demonstrated that *CDH1* plays a key role in both carcinogenesis and craniofacial morphogenesis. This reinforces the potential connection between *CDH1* variants, cancer susceptibility, and the risk of orofacial clefts.

CONCLUSION

The analysis reveals that nonsynonymous variants in the *CDH1* gene are associated with both orofacial clefts and cancer, although these associations lack functional validation. Importantly, no single variant was found to be concurrently associated with both phenotypes.