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SNPs in genes involved in sweet taste perception and intake (SweetG) are associated with Primary Ciliary Dyskinesia (PCD) and related phenotypes

Author Block: Romina Ruberto[1], Gioia Piatti[2,3], Silvia Camarda[4], Giuseppe Giovanni Nardone[4], Alessandro Pecori[1], Aurora Santin[1,4], Paola Tesolin[1,4], Elisabetta Tassin[5], Beatrice Spedicati[1,4], Giorgia Girotto[1,4], Maria Pina Concas[1].

[1]Institute for Maternal and Child Health, I.R.C.C.S. "Burlo Garofolo", Trieste, Italy, [2]Department of Pathophysiology and Transplantation, University of Milan, Milan, Italy, [3]Unit of Bronchopneumology, Fondazione I.R.C.C.S. Ca' Granda Ospedale Maggiore Policlinico, Milan, Italy, [4]Department of Medicine, Surgery and Health Science, University of Trieste, Trieste, Italy, [5]Department of Life Sciences, University of Trieste, Trieste, Italy.

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Background: PCD is a congenital motile ciliopathy caused by impaired mucociliary clearance, characterized by recurrent respiratory infections affecting both the upper and lower airways. Many genes involved in taste perception pathways are expressed in extra oral tissues and have recently emerged as regulators of airway immune responses. The aims of this study were to determine if SNPs in SweetG are more/less frequent in PCD patients, and verify their association with PCD clinical characteristics. **Material and Methods:** A list of seven SNPs in six SweetG have been tested for differences in allele frequency between PCD patients and gnomAD v4.1.0 using binomial test. Logistic and linear regression models have been performed to study the association between SNPs and PCD patients' clinical features. **Results:** A cohort of 34 PCD patients (6-69y, 56% female) was included in the study. The minor allele of rs5415 (*SLC2A4* gene) was less frequent in PCD patients than gnomAD ($p<0.05$). As regard PCD patients' clinical features, we found that rs5415 was

associated with a greater presence of chronic rhinosinusitis ($p<0.05$). In addition, rs7534618 (*TAS1R2* gene) was associated with the presence of situs inversus, and rs17457384 (*GABRB2* gene) was associated with a decreased respiratory function (FEV_1). **Conclusion:**

This study shows association between SweetG and specific PCD clinical features. The significance of these findings is not yet fully understood; however, this represents a promising area of research to enhance our understanding of PCD and elucidate the genetic basis of respiratory infections associated with this disease.

Author Disclosure Information:

R. Ruberto: None. **G. Piatti:** None. **S. Camarda:** None. **G. Nardone:** None. **A. Pecori:** None. **A. Santin:** None. **P. Tesolin:** None. **E. Tassin:** None. **B. Spedicati:** None. **G. Girotto:** None. **M. Concas:** None.

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