▲ I G E N E T I C S

Dental Report

Sample Details Sample ID: Test ID: A1 D01 Specimen Type: Buccal Collection Date: Received Date:

Ordering Clinician Clinician Name Phone: Email: Practice:

Your Results

Your Details

Full Name:

Age:

Sex: Patient ID:

Periodontal Disease: High Risk



The combination of genetic markers detected suggests that your baseline risk of developing periodontal disease is higher than the average person in the population.

Out of every 100 people, your risk is higher than 95 others.

Although your genetic risk is increased based on this polygenic risk score algorithm, this result does not mean that you have periodontal disease or that you are certain to develop it. You may wish to consider a proactive approach to oral care.

* Please note that a score within the 95th percentile does not mean that you have an 95% chance of developing periodontal disease.

Cavities: Low Risk



Relative Polygenic Risk Score Distribution

The combination of genetic markers detected suggests that your baseline risk of developing cavities is lower compared to the average person in the population.

Out of every 100 people, your risk is lower than 95 others.

Although your genetic risk is decreased based on this polygenic risk score algorithm, it is still possible to develop dental caries based on other lifestyle and clinical factors.

* Please note that a score within the 5th percentile does not mean that you have ar 5% chance of developing cavities

These results are not intended to be diagnostic, nor are they deterministic. See below for a more detailed explanation of these results and what it means for you.

Ai Genetics Holdings 1180 Aerowood Drive 2nd floor, Mississauga, ON L4W 1Y5 (905) 624-4363(GENE) support@aigenetics.ca

Sample Details Sample ID: Test ID: AI_D01 Specimen Type: Buccal Collection Date: Received Date: Ordering Clinician Clinician Name: Phone: Email: Practice:

Detailed Summary

Periodontal Disease

 Periodontal disease (PD) can range in severity from gingivitis to severe gum disease and tooth loss. This disease affects nearly 50% of adults over 30 in North America; with severe disease affecting approximately 11% of the global population^{1,2}. Severe PD is the main cause of tooth loss and is the most common oral condition in the human population¹.

Dental Cavities

Cavities (caries) impact both pediatric and adult populations. Approximately 26% of adults with cavities have untreated tooth decay³. Early childhood caries (ECC) impacts 1-12% of the pediatric population in developed countries. ECC can have detrimental consequences including pain, abscess formation, as well as difficulty eating and swallowing⁴.

Explanation of Results

- Genetics is one of many factors that may impact an individual's risk for developing periodontal disease and cavities, however, many other factors are involved including age, diet, oral hygiene routine, smoking, ethnicity, diabetes and the natural landscape of bacteria in the mouth⁵.
- There is evidence that some genetic markers are associated with a higher risk to develop periodontal disease and cavities while others are associated with a lower risk. People usually carry a combination of both⁵.
- A relative polygenic risk score (R-PRS) is a statistical way of combining genetic risk based on multiple genetic markers to determine a person's overall genetic risk relative to a reference population database⁶.
- This result does not mean that you have periodontal disease or cavities, or that you are certain to develop it, however you may wish to consider a proactive approach to oral care.

Next Steps

- Discuss these results with your dental healthcare provider so you can work together to develop an oral hygiene care plan that may help to reduce your risk of developing periodontal disease and cavities.
- For more information on best oral hygiene practices, consult the Canadian Dental Association webpage: <u>https://www.cda-adc.ca/en/oral_health/index.asp</u>

Sample Details Sample ID: Test ID: AI_D01 Specimen Type: Buccal Collection Date: Received Date: Ordering Clinician Clinician Name: Phone: Email: Practice:

Test Methodology

Patient DNA is genotyped using the Agena Bioscience MassARRAY® System. Up to a total of 38 genetic markers, passing the analytical quality control metrics, are evaluated in this test and used in subsequent R-PRS computation. The R-PRS development was performed using PLINK, a software tool for genomic analyses⁷. Separate polygenic risk scores are calculated for periodontal disease and dental cavity risk. Individual risk categories (low, intermediate-low, average, intermediate-high, or high risk) are determined by comparing the R-PRS of the patient to a normal distribution graph that is representative of the scores observed across the general population using a large ethnically diverse control cohort. The polygenic risk score is therefore intended to be relative to the general population.

Description of R-PRS Risk Categories

Risk Category	Percentile
Low	5 th
Intermediate-Low	15 th
Average	50 th (range from 15 th to 85 th)
Intermediate-High	85 th
High	95th

Disclaimer:

The genetic markers included in this test have evidence for association with increased risk for periodontal disease and cavities based on a review of the literature and have therefore been selected for inclusion in the development of the polygenic scores. While some of these markers are located within genes that are associated with rare genetic conditions, the results of this test are reported within the context of periodontal disease and cavities only.

Test Limitations

Molecular tests are highly accurate, however, rare diagnostic errors may occur that interfere with analysis. Sources of these errors include sample mix-up, trace contamination, and other unforeseen technical errors. Laboratory errors occur at a rate of approximately 0.05% in patient samples⁸. This test does not account for all possible genetic markers associated with periodontal disease and cavities. Polygenic scores are not to intended to be used as diagnostic tools, nor are they meant to be deterministic. People with high polygenic scores may not develop periodontal disease or cavities, while people with low scores may develop these conditions. Genetics is one of many factors that may impact risk to develop periodontal disease and cavities including but not limited to age, diet, oral hygiene routine, smoking, ethnicity, diabetes, and the natural landscape of bacteria in your mouth⁵. The R-PRS does not take these other factors into account.

Regulatory Disclosures

This test was developed, validated, and its performance characteristics determined by Ai Genetics Laboratories.

References

- Eke, P. I., Dye, B. A., Wei, L., Thornton-Evans, G. O., & Genco, R. J. (2012). Prevalence of periodontitis in adults in the United States: 2009 and 2010. Journal of Dental Research, 91(10), 914–920. <u>https://doi.org/10.1177/0022034512457373</u>
- GBD 2016 Disease and Injury Incidence and Prevalence Collaborators. (2017). Global, regional, and national incidence, prevalence, and years lived with disability for 328 diseases and injuries for 195 countries, 1990-2016: a systematic analysis for the Global Burden of Disease Study 2016. *Lancet* (*London, England*), 390(10100), 1211–1259. <u>https://doi.org/10.1016/S0140-6736(17)32154-2</u>
- 3. National Institute of Dental and Craniofacial Research. (2018). *Dental caries (tooth decay) in adults (Age 20 to 64)*. https://www.nidcr.nih.gov/research/data-statistics/dental-caries/adolescents
- Anil, S., & Anand, P. S. (2017). Early childhood caries: Prevalence, risk factors, and prevention. Frontiers in Pediatrics, 5, 157. https://doi.org/10.3389/fped.2017.00157
- 5. Patel, A. B., Matthews, D. C., and Ghiabi, E. (2016). Practice profile of periodontitis in Canada: A national survey. *Journal of the Canadian Dental Association; 82,* g5.
- Choi, S. W., Mak, T. S., & O'Reilly, P. F. (2020). Tutorial: a guide to performing polygenic risk score analyses. *Nature Protocols*, 15(9), 2759–2772. https://doi.org/10.1038/s41596-020-0353-1
- Purcell, S., Neale, B., Todd-Brown, K., Thomas, L., Ferreira, M. A. R., Bender, D., Maller, J., Sklar, P., de Bakker, P. I. W., Daly, M. J. & Sham, P. C. (2007). PLINK: a toolset for whole-genome association and population-based linkage analysis. *American Journal of Human Genetics*, 81(3), 559–575. https://doi.org/10.1086/519795
- 8. Bonini, P., Plebani, M., Ceriotti, F., & Rubboli, F. (2002). Errors in laboratory medicine. Clinical Chemistry, 48(5), 691-698.

Signature

Priscillia Quan, MD

Ai Genetics Holdings 1180 Aerowood Drive 2nd floor, Mississauga, ON L4W 1Y5 (905) 624-4363(GENE) <u>support@aigenetics.ca</u>