



## Saliva Collection is not a Universal Sample Collection Tool: 20% - 46% of the Population Suffers from Dry Mouth

Not everybody can produce enough saliva: Collecting 1-2 mL of Saliva within 30 minutes (as required by most saliva collection kits) can be very challenging for a significant portion of the population, especially children and elderly. It is well documented that as much as **20% - 46%**<sup>1</sup> of the population suffers from a condition known as dry mouth (xerostomia), which results from minimal or no saliva production.

- Typical causes of dry mouth:
  - Common, over the counter drugs such as allergy medication and cough syrups
  - Prescription medications for hypertension or depression.
  - Chronic diseases such as Sjögren's, sarcoidosis, hepatitis C, diabetes, or depression
  - Poor diet
  - Age
  - Heredity
- Saliva (and therefore DNA/genes) can be exchanged between individuals during mouth-to-mouth contact:

### Lovers Swap Genes While Kissing: DNA Lingers in the Mouth Even After a Light Peck

<http://www.medicaldaily.com/lovers-swap-genes-while-kissing-dna-lingers-mouth-even-after-light-peck-244393>

- Saliva is a complex matrix composed from water, mucus, white blood cells, some epithelial cells, digestive enzymes, and bacteria. The source of gDNA in saliva mainly comes from white blood cells<sup>3</sup>, which are usually lacking in patients undergoing chemotherapy (leukopenia<sup>3</sup>). This results in low gDNA yields making saliva an improper sample type for molecular analysis of chemotherapy effectiveness.
- Saliva contains high bacterial content and food particles that increase sample failure and hinder sample processing for applications such as genotyping, DNA sequencing, drug testing, or protein marker screening. This is often why service providers ask for additional samples, however the compliance rate is only about 50%. Researchers have utilized chemistries from companies like New England Biolabs to selectively deplete high bacterial DNA contamination from saliva to enhance their human DNA sequencing coverage, but this step adds time and cost to the workflow.

### Why is there a need for a universal sample collection method?

The use of Saliva collection devices such as spit tubes, drool capture, gauze wrapped cotton balls, mouth wash, or foam based swab saliva collectors as the only sample collection tool for genealogy (ancestry), laboratory developed tests or population studies is providing us with just 54% - 80% of the genomics picture for the community or population of interest. This means as much as 20% - 46% of the picture is missing, leaving quite a bit of room for speculation...

This is exactly why over 9 years was invested in developing the iSWAB non-invasive sample concentration and transport technology for collection of buccal (cheek) cells. iSWAB utilizes standard, off the shelf swabs made of cotton or other materials as the collection tool which is an established method familiar to most people. Another

advantage of using swabs is that they allow selective collection of buccal cells, the major component of the inside of the cheek<sup>4</sup>. It is well documented that buccal cells are a more informative surrogate tissue than blood for epigenome-wide association studies<sup>5</sup>. Studies have shown that the genetic material found in buccal cells (as opposed to white blood cells) is more similar to other major body tissues and therefore provides more relevant and standardized information across different populations.

The **iSWAB** technology is suitable for any population segment, significantly reduces bacterial DNA contamination with no extra steps involved, and produces robust yields of high quality gDNA. Our proprietary iSWAB buffer selectively lyses mammalian cells and stabilizes the gDNA for room temperature transport and long-term storage while leaving the bacterial DNA permanently inaccessible<sup>6,7</sup>. iSWAB works by allowing collected buccal cells to be scraped off the swabs and suspended in our buffer. After collection, the swabs are discarded and do not accompany the sample. This feature gives the added advantage of making sample processing faster, easier, and automation friendly.

In conclusion, gaining access to the majority of participants regardless of age or physical/mental status allows us to better understand the genetic picture and underlying disease factors for a specific population or community. Having a universal non-invasive method to collect samples other than saliva is critical to achieve this objective more precisely.

### **References:**

1. Leo M. Sreebny and Arjan Vissink, **Dry Mouth, the Malevolent Symptom** (Hoboken, NJ: Wiley-Blackwell, 2010), 12, fig. 1.2.2.
2. <http://www.medicaldaily.com/lovers-swap-genes-while-kissing-dna-lingers-mouth-even-after-light-peck-244393>
3. Sepúlveda E, Rojas IG, Brethauer U, Maulén NP, Muñoz M, Kirsten L, Oñate A, Fernández E, Le Fort P & Rojas J. **Effect of white cell counts on the presence of human herpes simplex virus type-1 in saliva of pediatric oncology patients**. Oral Surg Oral Med Oral Pathol Oral Radiol Endod. 2008 May;105(5):583-8.
4. Esther R. Berko, Masako Suzuki, Faygel Beren, Christophe Lemetre, Christine M. Alaimo, R. Brent Calder, Karen Ballaban-Gil, Batya Gounder, Kaylee Kampf, Jill Kirschen, Shahina B. Maqbool, Zeineen Momin, David M. Reynolds, Natalie Russo, Lisa Shulman, Edyta Stasiak, Jessica Tozour, Maria Valicenti-McDermott, Shenglong Wang, Brett S. Abrahams, Joseph Hargitai, Dov Inbar, Zhengdong Zhang, Joseph D. Buxbaum, Sophie Molholm, John J. Foxe, Robert W. Marion, Adam Auton, & John M. Greally Lowe R, Gemma C, Beyan H, Hawa MI, Bazeos A, Leslie RD, Montpetit A, Rakyen VK, & Ramagopalan SV. **Mosaic Epigenetic Dysregulation of Ectodermal Cells in Autism Spectrum Disorder**. PLoS Genet 10(5): e1004402.
5. Lowe R, Gemma C, Beyan H, Hawa MI, Bazeos A, Leslie RD, Montpetit A, Rakyen VK, & Ramagopalan SV. **Buccals are likely to be a more informative surrogate tissue than blood for epigenome-wide association studies**. Epigenetics. 2013 Apr;8(4):445-54
6. Kiranmai Durvasula, Jeff Roeder, Travis Butts, & Bassam El-Fahmawi. **Comprehensive, high through-put workflow for automated gDNA isolation from iSWAB oral samples**. Poster Presentation at ASHG-2016, PgmNr 1011/F
7. Andrew D. Johnston, Bassam El-Fahmawi, & John M. Greally. **Selective Lysis Enables Effective Whole Genome Bisulphite Sequencing of Buccal Epithelial Samples**. Epigenomics 2016, Poster Presentation.

