

1. Gene content, phage cycle regulation model and prophage inactivation disclosed by prophage genomics in the Helicobacter pylori Genome Project

Contenido genético, modelo de regulación del ciclo de los fagos e inactivación de los profagos revelados por la genómica de los profagos en el Proyecto Genoma de Helicobacter pylori

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ABSTRACTO: Prophages can have major clinical implications through their ability to change pathogenic bacterial traits. There is limited understanding of the prophage role in ecological, evolutionary, adaptive processes and pathogenicity of Helicobacter pylori, a widespread bacterium causally associated with gastric cancer. Inferring the exact prophage genomic location and completeness requires complete genomes. The international Helicobacter pylori Genome Project (HpGP) dataset comprises 1011 H. pylori complete clinical genomes enriched with epigenetic data. We thoroughly evaluated the H. pylori prophage genomic content in the HpGP dataset. We investigated population evolutionary dynamics through phylogenetic and pangenome analyses. Additionally, we identified genome rearrangements and assessed the impact of prophage presence on bacterial gene disruption and methylome. We found that 29.5% (298) of the HpGP genomes contain prophages, of which only 32.2% (96) were complete, minimizing the burden of prophage carriage. The prevalence of H. pylori prophage sequences was variable by geography and ancestry, but not by disease status of the human host. Prophage insertion occasionally results in gene disruption that can change the global bacterial epigenome. Gene function prediction allowed the development of the first model for lysogenic-lytic cycle regulation in H. pylori. We have disclosed new prophage inactivation mechanisms that appear to occur by genome rearrangement, merger with other mobile elements, and pseudogene accumulation. Our analysis provides a comprehensive framework for H. pylori prophage biological and genomics, offering insights into lysogeny regulation and bacterial adaptation to prophages.

2. Feasibility of an environmental scan-based approach to collecting information about factors impacting cancer genetics services in Latin American countries

Viabilidad de un enfoque basado en el análisis ambiental para recopilar información sobre los factores que afectan los servicios de genética del cáncer en países de América Latina

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ABSTRACTO: Objective: Clinical cancer genetics services are expanding globally, but national policy and health care systems influence availability and implementation. Understanding the environmental factors within a country is required to appropriately implement, adapt, and evaluate cancer genetics service delivery models. An environmental scan (ES) is an approach used in business, public health, health care and other sectors to collect information about an environment or system for strategic decision making and program planning. An ES has been previously used to assess cancer genetics clinic-level factors to inform quality improvement efforts in the United States. We assessed the feasibility of using an ES to collect information about factors that may influence cancer genetics service delivery in the outer-most socio-ecological model environmental levels (policy, national agencies, healthcare systems, cultural considerations) in three Latin American countries. Methods: Oncology and Genetics care team members at three participating sites used publicly available sources and personal experiences to complete a data collection form (DCF) that included questions about subtopics: laws and policies, relevant agencies and regulations, health care systems and insurance, and cultural considerations. Time to complete the DCF and DCF completeness were used to measure ES feasibility. Results: Participating sites completed the DCF in 3 months, and most questions (average, 87.0%) were answered. Questions in the cultural considerations subtopic had the fewest answers (average, 77.8%). Conclusions: Overall, the ES was feasible and identified a lack of published literature related to cultural considerations impacting health care and genetics services uptake in Latin America. Environmental factors impact cancer genetics services, and identification of these factors will facilitate future collaborative research and genetics service delivery dissemination efforts.

3. Cancer genetic counseling in Chile: Addressing barriers, confronting challenges, and seizing opportunities in an underserved Latin American Community

Consejería genética sobre el cáncer en Chile: Abordando barreras, enfrentando desafíos y aprovechando oportunidades en una comunidad latina desatendida

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ABSTRACTO: Purpose: Despite the rapid advancements in genomics and the enactment of a new cancer law in Chile, the implementation of cancer genetic counseling continues to face significant challenges because of limited resources and infrastructure. Methods: We conducted a survey targeting health care providers who offer genetic counseling to patients with cancer and possess training in genetics and counseling. Additionally, we distributed a separate survey to high-risk patients associated with an advocacy group to gather insights on their perceptions of and experiences with cancer genetic counseling. Results: Among the surveyed providers, 21% were nonmedical professionals who developed their skills through postgraduate continuing education programs. Germline testing was not performed in 47% of cases. Among the participants, 37% considered

genetic counseling important for understanding the cause of their cancer, 25% valued knowing their risk of developing future tumors, and 33% believed it would benefit their current cancer treatment. Just over half of the patients (54%) had access to genetic counseling. Among those that received genetic counseling, 85% found it beneficial. Conclusion: In Chile, barriers to genetic counseling persist, particularly in rural areas and because of a shortage of trained professionals. Public policies recognizing genetic counseling's importance are crucial, along with expanding training and infrastructure. Understanding patient perceptions and increasing the number of trained genetic counseling into cancer care, educating clinicians, and advocating for increased access are key steps for enhancing cancer treatment effectiveness in Chile.