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SPECIAL ARTICLE

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

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ABSTRACT

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.

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Introduction

Cancer genetic counseling is a critical field in the fight against cancer, and in Latin America, the diversity of ethnic ancestries adds an additional layer of complexity and relevance to this field.[1] One area in which ethnic diversity has a significantly impact is in genetic predisposition to cancer. Access to genetic counseling in Latin America varies by country because of differences in health care infrastructure and policies. Argentina and Brazil have more widespread access with specialized centers and trained professionals in clinical genetics, along with public policies supporting genetic research and counseling services.[2–7]

In Chile, access to genetic counseling may be limited because of factors such as lack of resources and underdeveloped health care infrastructure resulting in a shortage of professionals trained in clinical genetics and a lack of specialized centers for genetic counseling.[8,9] In August 2020, Chile enacted the National Cancer Law 21.258,[10] aiming to enhance the prevention, diagnosis, treatment, and follow-up care for patients with cancer. Article 90 of this law mandates medical teams to provide genetic counseling to patients with cancer with high-risk factors. However, implementation of this provision is still pending.

Limited access to genetic counseling is also influenced by the fact that marginalized and rural populations in Chile may have difficulty accessing quality health care, including genetic counseling. This is due to a lack of education about genetics and health, which leads to low utilization of genetic counseling services and hinders efforts to make it universally available.[11–13]

Another key factor affecting access to genetic counseling in Latin America is the legislation and policies concerning genetics and genetic information privacy. Some countries have strict laws governing genetic testing and the management of patients' genetic information, which can affect the availability and accessibility of genetic counseling services.[14,15] A common concern during the genetic counseling process is patients' fear of their genetic data being disclosed to private insurance companies. Therefore, it is crucial to provide patients with reassurance that their clinical records are safeguarded as part of the genetic counseling process.[16]

Despite challenges, there have been significant advances in genetic counseling in Latin America. Countries are strengthening genetics programs, expanding specialized centers, and promoting equity in access to genetic health services.[17] International collaboration and the efforts of nongovernmental organizations have also contributed to improving access to genetics and genetic counseling in Latin America.[7,18] Organizations such as the Pan American Health Organization and the Latin American Association of Genetics have been working closely with governments and health institutions in the region to strengthen genetic counseling programs and raise awareness about the importance of genetics in public health.

Although cancer genetic counseling in Chile has made progress, it still faces challenges. There are gaps in availability, professional training, and socioeconomic and cultural barriers that persist. Access is difficult in rural areas, and there is a shortage of trained professionals. Overcoming these challenges requires expanding training, improving infrastructure, reducing barriers, and promoting policies to ensure equitable access to cancer genetics counseling nationwide. Recently, and in an effort carried out by the Chilean Hereditary Cancer Group, we showed together with a group of experts from Latin America and Spain, the greatest challenges, and opportunities regarding genetic counseling in Chile.[19]

Understanding how patients perceive cancer genetic counseling is also crucial. In countries where genetic counseling is universally recognized, patient perceptions may differ significantly from those where it is not.[20] Recognizing genetic counseling as an essential service for patients with cancer could guide public policies in Chile to establish it as a crucial profession within the health system.

At the same time, it is essential to determine the number of health care professionals who provide genetic counseling, their training, and areas of specialization to improve health care and address hereditary diseases. A study conducted in 2013 revealed that there was only 1 genetic counselor and 28 clinical geneticists in a country with a population of over 16 million.[8] The clinical geneticists primarily focused on pediatric dysmorphology conditions, and cancer genetics conditions were not a priority in health care.

Except for Brazil, which offers a master's degree in genetic counseling in Portuguese, most Spanish-speaking Latin American countries lack specialized training programs in this field. In Chile, genetic counseling remains unregulated. Article 90 of Decree 42, part of the 2020 Cancer Law 21.258,[10] understands genetic counseling in cancer as providing information regarding cancer development risks, the probability of hereditary transmission, and strategies for early diagnosis and prevention, delivered by a health care professional. Genetic counseling is the process of helping people understand and adapt to the medical, psychological, and familial implications of genetic contributions to disease.[21,22] However, the law does not specify the formal training requirements for genetic counselors and narrowly focuses on providing information, neglecting the essential psychosocial aspects of the counseling process.

Therefore, there is an urgent need to monitor the number of genetic counselors, understand their training and specializations, and coordinate their efforts with other health care providers to enhance the quality and effectiveness of services. Finally, it is imperative to evaluate patients' needs based on socio-cultural characteristics along the Chilean ancestral diversity.

To gather this information, we asked health care providers and patients with cancer to participate in a brief anonymous survey. Our goal was to improve our

understanding of genetic counseling practices in cancer genetics. We believe that the survey results offer valuable insights into the perception and utilization of cancer genetic counseling, as well as identify necessary improvements to enhance its accessibility and effectiveness within the public health care system. The collected information will contribute to the future development of cancer genetic counseling services and underscore the national demand for genetic counseling in other critical areas that also require attention and resources.

Materials and Methods

An online survey was designed to collect data from health care providers and patients with cancer regarding genetic counseling services. We focused on obtaining information that was most relevant according to the Chilean reality. Because of the lack of programs and implementation of genetic counseling, the application of tools validated in other countries does not fit the current situation in Chile. Therefore, the survey collected information on demographic factors, professional training, and experiential perceptions to evaluate the current state of genetic counseling. The objectives of this data collection were to identify deficiencies in knowledge, determine the need for additional training, and assess the levels of awareness and attitudes toward genetic counseling among both health care providers and patients.

Survey for genetic counseling professionals

The survey targeting genetic counseling professionals sought to collect detailed information on their professional backgrounds. It included queries regarding demographic characteristics, duration and extent of training, dedication to cancer genetic counseling, professional experience, and perceptions of the challenges encountered in the field.

Survey for patients with cancer

The survey was distributed to patients with cancer in Copiapó, a city in northern Chile. Most participants lacked private insurance and were covered by the public health care system. They were also members of the advocacy group Women's Breast Cancer Society. The survey questions focused on the accessibility, usefulness, and perceptions of genetic services.

Institutional Review Board

Institutional Review Board approval was not required because participation in this study was voluntary, and informed consent was implied by completion of the anonymous online survey.

Results

The survey included 20 genetic counseling professionals (Table 1), 85% of whom were between 30 and 50 years old. Additionally, 95% of the participants had education in genetic counseling and oncology, and 90% of them were physicians. Most of the participants were clinical geneticists working in public and private hospitals, mainly focusing on pediatric genetics diagnosis and occasionally on cancer risk assessment. These clinical geneticists were educated through the only 2 clinical genetics residency programs in Chile, based in Santiago, producing an average of 2 graduates per year. Seventy-nine percent of the surveyed professionals have acquired genetic counseling skills during their clinical genetics' residency, whereas 21% were nonmedical professionals who obtained postgraduate genetic counseling skills through specific genetic counseling continuing education programs.

According to the study, germline testing was not ordered in 47% of cases. The primary challenge faced by genetic counseling professionals in Chile were the lack of coverage for clinical consultations and hereditary assessments by the public health system (35%), as well as limited access to genetic counseling and germline diagnosis.

Patients' survey

A total of 69 patients (Table 2) who have received diagnosis for triple-negative breast cancer (TNBC) and are associated with a nonprofit organization for cancer survivors took part in the survey. These groups are now more frequently resorting to legal action in diagnostic processes, pushing for the country to fund these cases.

Among the participants, 37% found genetic counseling important for understanding the cause of their cancer, whereas 25% valued knowing their risk of developing future tumors, and 33% believed that it would be beneficial for their current cancer treatment.

Just over half of the patients (54%) had access to genetic counseling, and many of these consultations were provided free of charge by the Chilean Hereditary Cancer Group. Additionally, only 51% of patients were 45 years old or younger. Fifty-nine percent of the patients stated that clinicians had referred them for genetic counseling. All of the patients (100%) agreed on the importance of knowing whether they had a positive carrier status for any pathogenic germline genetic variant. Among those that received genetic counseling, 85% found it beneficial. Additionally, almost all, 97%, reported advantages in their treatment after consulting with a genetic counselor.

Discussion

In Chile, cancer genetic counseling is a rapidly evolving field within oncology, driven by advancements in precision

Table 1 Results of survey of clinical geneticists

Age	20-30	5%
	30-40	45%
	40-50	40%
	>60	10%
Gender	Male	40%
	Female	60%
Education in other areas of GC than cancer	Yes	95%
	No	5%
Type of education	>2 years of masters	20%
	Diploma	15%
	Short-term course	20%
	Linked to medicine or nurse	40%
	Other	5%
Undergraduate studies	Physician	90%
	Nurse	5%
	Biologist	5%
Postgraduate studies other than GC	Medical specialty	79%
	Master	21%
Place of education in GC	Chile	85%
	Abroad	15%
Dedication to cancer GC	Mostly	45%
	General GC	55%
Years of practice	0-10	65%
	10-15	5%
	>15	30%
Frequency of genetic testing request	0%-10%	47%
	10%-25%	16%
	25%-50%	5%
	50%-70%	11%
	>70%	21%
Main challenge for GC in Chile	Lack of reconnaissance	20%
	Lack of access	20%
	Lack of coverage	35%
	Lack of education in GC	15%
	Lack of regulation	10%

GC, genetic counseling.

Table 2 Results of survey of patients

GC is useful to know	The reason of my cancer	37%
	I will develop cancer	25%
	How to treat my cancer	33%
	Do not know	5%
Did I have access to GC	Yes	54%
	No	46%
Age	18-45	51%
	>45	49%
Institution where the diagnosis was established	Public	61%
	Private	39%
Could GC be useful to my relatives?	Yes	97%
	No	0%
	Do not know	3%
Would I like to know if my cancer is hereditary?	Yes	100%
	No	0%
Relatives with cancer	Yes	70%
	No	30%
Could GC facilitate the cancer treatment journey?	Yes	97%
	No	3%
Was GC offered to me by the clinician?	Yes	59%
	No	41%
In case of have received GC, was it useful?	Yes	85%
	No	15%

GC, genetic counseling.

medicine.[23] Access to genetic counseling may be limited because of factors such as lack of resources and underdeveloped health care infrastructure, resulting in a shortage of professionals trained in clinical genetics and a lack of specialized centers for genetic counseling. In August 2020, Chile enacted the National Cancer Law 21.258,[10] aiming to enhance the prevention, diagnosis, treatment, and follow-up care for patients with cancer. Article 90 of this law mandates medical teams to provide genetic counseling to patients with cancer with high-risk factors. However, implementation of this provision is still pending.

In our study, we focused specifically on patients with TNBC, a subgroup for whom genetic counseling is particularly relevant because of the higher likelihood of hereditary risk, distinguishing them from both the general cancer population and other breast patients with cancer. Although TNBC offers a valuable lens for examining broader issues related to access to genetic counseling, we acknowledge that some of our findings—such as the 54% referral rate—may not be directly applicable to all patients with cancer, particularly those with lower hereditary risks. In the general cancer population, in which many individuals lack a family history or elevated genetic risk, referral rates for genetic counseling would understandably be lower. This finding highlights significant gaps in access, even within this high-risk group, and points to broader systemic issues related to the availability and accessibility of genetic counseling in cancer care.

The survey for genetic counseling professionals revealed that most respondents were middle-aged clinical geneticists with MD degrees, primarily working in public and private hospitals. Their practice was mainly on pediatric genetic diagnosis, with occasional involvement in cancer risk assessment. These clinical geneticists were trained through the only 2 genetics residency programs in Chile, both located in Chile's capital Santiago, producing an average of just 2 graduates per year. Currently, Chile lacks a formal program in genetic counseling, leaving clinical geneticists to address the general needs for genetic counseling. However, these professionals do not have access to training programs that meet the genetic counseling standards established in the

United States or Europe.[24] This underscores the significant shortage of experienced cancer genetic counselors in Chile, posing a major challenge to advancing educational initiatives in this field.

Close to 80% of the surveyed professionals have acquired genetic counseling skills during their clinical genetics' residency, and the rest obtained postgraduate genetic counseling skills through continuing education programs, short-term courses that did not offer or required practice evaluation. The absence of formal, supervised genetic counseling programs has fostered the misconception that anyone who completes these brief courses can consider themselves genetic counselors, despite lacking proper supervision or certification.

Only 1 genetic counselor (coauthor of current publication) pursued a master's degree from an established program in the United States. This highlights the urgent need for a certified program in the region. Such a program should involve collaboration with other Spanish-speaking countries with similar needs, such as Argentina, Peru, Uruguay, Bolivia, etc. Undergraduate exposure to genetic counseling can increase interest, especially among underrepresented populations who are currently underrepresented in the field. Although most genetic counselors are White and female, students from minority groups are equally likely to consider this career if they are introduced to it.[25] However, these students typically have less exposure to genetic counseling. Research suggests that increasing exposure among undergraduates may boost awareness and interest in the field among minority students.[26] Sixty-five percent of specialists have less than 10 years of practice, and their dedication is not focused on cancer genetic counseling. This could be partially explained by the fact that several clinical geneticists, who were educated by the abovementioned program, are required to work and repay the years of education they received through pediatric genetics mostly.

Efforts should prioritize educating clinicians on recognizing red flags in patients who meet high-risk criteria and to distinguish and order germline and somatic genetic tests.[27] This approach would minimize confusion and facilitate accurate referrals. To ensure that nongenetics health care providers are adequately trained, a structured system of continuing education in genetic counseling is crucial.[28,29] This system would certify that these professionals receive the necessary education, helping to alleviate the shortage of human resources in the field. In Chile, 2 online certificate diploma courses currently offer specialized training in genetic counseling for health care providers. The first, offered by the Catholic University of the Maule Region, focuses on cancer genetic counseling, emphasizing the clinical and genetic aspects of hereditary tumors. The second, provided by Clínica Alemana Universidad del Desarrollo, offers a broader approach, with a diploma certificate in genetic counseling that covers hereditary conditions across various areas of health care.

The incorporation of Next-Generation Sequencing (NGS) into patient management presents several challenges for health care providers, including interpreting results, deciding their clinical relevance, and effectively communicating findings to patients and families.[30] These issues are particularly relevant in oncology, in which NGS is increasingly used to identify specific mutations and altered pathways for targeted therapies. Additionally, there is growing interest in using NGS germline assessment to detect cancer susceptibility syndromes, given its potential for enhancing cancer surveillance and risk reduction.[31] In Chile, there is no formal education that allows clinicians to learn and apply strategies regarding the ordering and interpretation of both germline and tumor assessment although the Chilean Society for Clinical Oncology has announced the launch of a National Molecular Tumor Board free of charge for oncologists belonging to the society.

Dick et al,[32] (2020) in Germany revealed crucial insights into physicians' understanding of genetics, cancer risk communication, and the clinical management of at-risk patients. Most respondents recognized the growing importance of genetic testing in oncology for treatment and prevention and used the German national criteria and checklists to screen patients for hereditary cancer risk.[32] However, studies indicate that only a small percentage of eligible patients undergo genetic testing, which is very similar to the results we found in Chile where only 54% of women diagnosed with TNBC had access to genetic counseling. This limitation is partly due to the shortage of genetic counselors and the limited genetic knowledge among clinicians in Chile as it was observed in Germany.

Blee et al[33] (2021) show that oncologists use varied and sometimes inconsistent terminology when describing molecular testing, with 18 different terms being used interchangeably. For instance, "molecular status" was used in place of "driver mutation," and "oncogene" was used for "biomarker." This inconsistency may contribute to patients' poor understanding of technical terms. Nearly half of the patients struggled to define "mutation," and just over half correctly defined "genetic testing." Terms such as "DNA" and "targeted therapy" were better understood, but a clinician's explanation did not necessarily improve patient comprehension. Chile has a vast territory along more than 5000 km with different climates, cultures, and ancestries. The heterogeneity of vocabulary is also present here and partially explained by the variety of professionals who deliver this genetic information. Historically, genetic counselors could become certified and practice with varying educational backgrounds. However, the field has evolved, and professional standards now require a master's degree in genetic counseling or a closely related field for certification and practice.[32] Neither in Chile nor in the rest of the Spanish-speaking countries in Latin America, as mentioned before, there are established master's degrees in genetic counseling. The application of a Latin American Grandfather's clause could facilitate the certification and homologation of proper genetic counselors, allowing genetic counselors who were already practicing before the implementation of these new educational requirements to

continue working without needing to obtain a master's degree.[34] To further ensure that these professionals meet the updated standards, an exam could be introduced as a mechanism for homologating their knowledge and competencies, validating their expertise while maintaining the integrity of the profession. This approach would help balance the introduction of new standards with the recognition of the experience and qualifications of those who were already established in the field fostering the beforementioned Chilean Cancer Law.

In relation to our patient survey, most of the respondents are affiliated with patient advocacy groups, which is becoming more prevalent in Chile. These groups work with political authorities and have an impact on the public health system as it has been seen in other countries.[35–37] The survey further highlighted diverse patient perspectives on the significance of genetic counseling in the context of their cancer journey. Notably, 37% of the respondents identified genetic counseling as important for understanding the cause of their cancer, indicating a significant demand for insights into the genetic factors underlying their diagnosis. Additionally, 25% of patients valued genetic counseling for its role in assessing their risk of developing future tumors, highlighting the importance of proactive health management. Furthermore, 33% believed that genetic counseling would be beneficial for informing their current cancer treatment, demonstrating its perceived value in tailoring therapeutic strategies to individual genetic profiles. Patients generally perceive cancer genetic counseling positively, appreciating the opportunity to gain a deeper understanding of their genetic risk factors, which aids in making informed health decisions.[38–40] The emotional support provided during counseling is particularly valued because the process can be anxiety inducing, especially when discussing potential cancer risks. Many patients feel empowered by the information they receive because it gives them greater control over their health through preventive measures, such as increased screening or lifestyle changes.[41] However, concerns about potential discrimination, particularly regarding insurance coverage or employment, are also common in Chile where private insurances might be the providers of germline assessments. Additionally, perceptions are influenced by the availability of genetic counseling services because we have limited access in some areas of the country causing frustration or a sense of being less informed about options.

Despite the recognized importance of genetic counseling in cancer care, only 54% of surveyed patients affected by cancer had access to these services. This limited access can be attributed to several factors.[3,16,39,42,43] One major issue is that physicians may not be adequately identifying patients at increased genetic risk, resulting in missed opportunities for referrals to genetic counseling. Additionally, both physicians and patients often lack a clear understanding of what genetic counseling involves and its potential benefits, leading to underutilization. Another significant barrier is the absence of health insurance coverage for genetic counseling services, making them financially inaccessible for many patients.[44,45] Furthermore, the availability of genetic counselors is highly concentrated in major metropolitan areas, leaving regional and rural communities underserved.[11,12] This centralization means that nongenetics professionals in these areas, who could otherwise provide preliminary guidance or referrals, are scarce or entirely absent, further limiting patient access to essential genetic services across Chile.

Even high-risk patients from low socioeconomic backgrounds were often not tested because of limited resources, including financial constraints and inadequate infrastructure. According to the results of the patients' survey, only 51% of patients were 45 years old or younger, and only 59% were referred to genetic counseling. However, if we consider consultations delivered through traditional means, this might lead to an overestimation of the services available, indicating that a significant proportion of older patients who could also be considered high risk might not have access to genetic counseling or germline genetic testing according to conventional clinical guidelines.[46] Furthermore, the lack of comprehensive epidemiological data on hereditary tumors in Chile prevents us from estimating the clinical and molecular characteristics of a diverse and underrepresented population. To ensure quality diagnosis and minimize patient distress, it is vital to automatically refer patients with suspected hereditary cancer to certified professionals. Essential steps in our country include increasing access to genetic counseling to all high-risk patients, even those over 45.

Hundred percent of the patients expressed the importance of knowing their carrier status, underscoring the high value they place on understanding their genetic risks. Additionally, 85% of respondents believed that genetic counseling is beneficial, and an overwhelming 97% felt it would positively affect their treatment. These findings clearly demonstrate that genetic counseling services play a crucial role in patient care, providing essential information that can significantly influence medical decisions and overall quality of life.[47] The strong support for these services highlights the need to make genetic counseling more widely available and accessible because it has the potential to make a profound difference in the lives facing cancer.

The absence of a national cancer registry, along with disparities in coverage and centralized oncology resources, led to the creation of a new National Cancer Plan in the 2018.[48,49] Nongovernmental organizations played a key role in advocating for an established National Cancer Law to serve as its legal framework, ensuring that the patient perspective is represented. Collaboration with clinical specialists, scientific societies, the government, and stakeholders were vital to address the complex challenge of cancer where it grants the right, for the first time, to genetic counseling and germline diagnosis.

Nongeneticists prescribe genetic tests, even without specific training in genetics.[50] This puts them in challenging

situations, requiring them to navigate not only the complexities of test relevance and interpretation but also the ethical and societal implications of testing. Communicating genetic status, obtaining consent, and managing the sharing of genetic information within families require skills many physicians lack. As genetic tests become more common across various medical fields, it is crucial to study how this shift affects practice, especially because many non-geneticists feel unprepared to handle genetic test results, and there is a lack of certified genetic counselors in Chile.

The data from the patient survey provide insight into the perspectives of clinicians who refer patients for cancer genetic counseling (59%). This emphasizes the importance of ensuring that any patient with suspected hereditary cancer is automatically referred to a certified professional to ensure a quality diagnosis.[51] This practice also helps prevent patients from experiencing distress and stress because they might otherwise misunderstand the genetic counselor's role

The surveys conducted with clinicians and patients had limitations. For clinicians, the small sample size, bias toward pediatric genetics expertise, and selection bias limited the representativeness and generalizability of the findings. The survey's focus on pediatric genetics likely skewed the results, neglecting the expertise required for cancer genetics counseling in adults. Additionally, nonresponder bias may have further distorted the outcomes. For patients, the survey faced limitations such as restricted access to participants, self-selection bias, and reliance on patient perceptions, which could misrepresent the actual utility of genetic counseling. These issues challenge the generalizability and completeness of the findings.

Conclusion

Cancer genetic counseling in Chile is advancing, but significant barriers remain that impede patient access to crucial services. Although the National Cancer Law 21.258 mandates genetic counseling for high-risk patients, its implementation is hindered by a lack of trained professionals and formal education in genetic counseling. Surveys show that genetic counseling professionals primarily focus on pediatric genetics, leading to disparities in cancer risk assessment services. Patients express a strong demand for genetic counseling, valuing its role in understanding their genetic risks and enhancing treatment outcomes. However, challenges such as inadequate physician training, limited insurance coverage, and resource centralization in urban areas restrict access for many. To improve the situation, a collaborative approach involving government, health care providers, and patient advocacy groups is essential. This should include implementing the National Cancer Law, establishing accredited training programs, expanding services to underserved areas, and raising awareness among health care professionals and patients. By taking these steps, Chile could ensure equitable access to genetic counseling and improve overall patient care and outcomes.

Data Availability

All data are shown within the manuscript.

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Ethics Declaration

Institutional Review Board approval was not required because participation in this study was voluntary, and informed consent was implied by completion of the anonymous online survey.

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Conflict of Interest

The authors declare no conflicts of interest.

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