アメリカの
遺伝カウンセラーの現状

遺伝カウンセラーの現状@アメリカ

- **状況**

  -全米 4,000人以上 (Stanford 約50人、UC Davis ９人)
  - Prenatal, Cancer, Cardiovascular, neuromascular などの専門領域がある
  - 病院（各領域、検査ｵｰﾀﾞｰ管理）, ラボ（commercial, in house), 大学, 企業（保険会社関係）, 行政など職場・職務は多岐に渡る
  - 遺伝カウンセラー単独の外来をもっていることが多い

- **養成コース**

  - 1969年～ , 現在39コース, 入学試験の倍率は**約10倍**
  - 統一されたシステムで国内全養成コースの入試を一括管理
    - 受験者：システム上で志望校に応募→書類選考→面接→
      志望校を順位付け（大学院側も受験者を順位付け）→合否決定
    - 絶対に１校にしか合格しない

  -受験者に求められるもの（足りないと面接までたどり着けない）
    - 陪席の経験, 遺伝カウンセラーアシスタントの経験（あるとかなり良い）,
      大学の成績, 基礎的な生物・遺伝関連の授業を事前履修

  - 生物学・心理学（学士）→Genetic Counselor Assistant→ 遺伝ｶｳﾝｾﾗｰｺｰｽ（修士）→ 認定試験受験

- **遺伝カウンセラーアシスタント**(or Administrator)

  - 事前に得た家系情報の整理やクライエント・患者への連絡, 検査キットの準備, 全般的な補佐を担当

遺伝カウンセラーの現状@アメリカ

- **認定試験（２種類）**
  - ABGC (American Board of Genetic Counseling)の試験に合格することによって認定遺伝カウンセラー(Certified Genetic Counselor(CGC))として認められる。5年更新。合格率約75％　(毎年2月と8月にコンピューター上で実施。全200問, 4時間。面接なし。当日に合否が判明。)
  - 各州独自の試験に合格するとLicensed Certified Genetic Counselor(LCGC)となる。カリフォルニアでは3年更新。

- **待遇**
  - 年収: 平均約810万円　新卒: 600-700万円　（アシスタント：350万円）
  - 経験に応じてGenetic Counselor I, II, IIIなど昇格制度を設けている病院もある

- **課題**
  - licenseがない州がある，勤務地変更でlicenseを取り直す必要
  - 勤め先による給料の格差
  - 白人女性が95%を占め, 多様性に欠ける
  - insurance matterに時間を割かなければならないことも多い

遺伝性腫瘍の
遺伝カウンセリング
@アメリカ（カリフォルニア）

遺伝カウンセリングの流れと担当者

流れ	担当者
遺伝カウンセリングの予約（院内紹介が主）	受付or 予約担当者
Webポータル経由で事前に家族歴の確認を依頼	遺伝カウンセラー
アシスタント等が家系図作成	
遺伝カウンセリング（家族歴の確認、疾患説明、検査説明など）、遺伝学的検査実施	遺伝カウンセラー
結果開示（電話or対面）	遺伝カウンセラー、医師（必要な場合）
Family letterの作成	
血縁者の遺伝学的検査、フォローアップ	遺伝カウンセラー
サーベイランス	医師

腫瘍領域のGC

- **状況**

  -スタンフォード大学では週に30〜60人のクライエント・患者が来院（年に2200人ほど）。遺伝カウンセラーが1人で年間350人ほどを担当。
  -基本的には遺伝カウンセラーのみで遺伝カウンセリング、遺伝学的検査の選択、結果開示、フォローアップを行い、必要に応じて医師が同席

- **遺伝学的検査**

  - 単一遺伝子検査は現在はほぼ使われていない
  - 複数の遺伝子を対象とするパネル検査が主流
  - 初回の来談で検査を実施することが多い
  - 遺伝学的検査を受ける目的で来る人が多いので、受検率は９５％以上
  - 万が一、受検者への結果開示が不可能になった場合のことを考慮し、代理に結果を開示してほしい人を事前に確認

- **その他**

  -遠方に住んでいる血縁者には近くの遺伝カウンセラーを紹介
  - サーベイランスに関しては、NCCNのガイドラインに沿って提示
  - 人種が多様のため、 Ashkenazi Jewishの祖先の有無を確認

腫瘍領域のGC

- 遺伝カウンセリング終了後、血縁者のリスクについてクライエント・患者が血縁者と情報共有ができるように、遺伝カウンセラーがFamily letterを作成。遺伝カウンセラーが血縁者と直接連絡をとることはなく、必ずクライエント本人を経由。家族の関係性によっては血縁者と共有されないこともある。

→次のスライドでFamily letter紹介

- 検査結果やFamily letterをWebポータル経由で送信

Family Letter(1/4)
～家族性腫瘍 遺伝学的検査の結果開示後～

Stanford HEALTH CARE

Stanford Cancer Genetics Clinic
900 Blake Wilbur Drive
Stanford, CA 94305-5820

2589 Samaritan Drive
San Jose, CA 95124-4100

Phone: 650-498-6000
http://cancer.stanford.edu/cancergenetics/

Licensed Genetic Counselors: [redacted], MS, LCGC · [redacted], MS, LCGC · [redacted] MS, LCGC
[redacted] MS, LCGC · [redacted], MS, LCGC [redacted] MS, LCGC · [redacted] MS, LCGC
[redacted], MD · [redacted] MD [redacted] MD · [redacted] MD · [redacted] PA · [redacted] NP

認定遺伝カウンセラー、医師等の名前

January [redacted] 2018

[redacted]

Dear [redacted]

パネル検査の対象遺伝子と検査結果(変異なし)

This letter summarizes results of the genetic testing that was ordered after your Stanford Cancer Genetics evaluation on [redacted] You reported a personal and/or family history suggestive of inherited cancer risk. You elected to proceed with the following genetic test:

- Multi-gene cancer risk panel through Invitae Laboratories. This included the following genes by sequencing and duplication/deletion analysis: *AKT1, ATM, BARD1, BRCA1, BRCA2, BRIP1, CDC73, CDH1, CHEK2, DICER1, EPCAM, FAM175A, FANCC, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, NF1, PALB2, PIK3CA, PMS2, POLD1, PTEN, RAD50, RAD51C, RAD51D, RINT1, SDHB, SDHD, SMARCA4, STK11, TP53, XRCC2*

Enclosed is a copy of your **negative** test result meaning no mutations were identified in the analyzed genes. We discussed at the time of your genetic counseling that testing for genes related to inherited cancer risk is not comprehensive. The possibility remains for a hereditary mutation in a gene that was not evaluated by this test, a small chance for a currently unidentifiable mutation within one of the genes analyzed, or the chance that the cancer in your personal and family history was not due to an inherited gene mutation.

Family Letter(2/4)
～家族性腫瘍 遺伝学的検査の結果開示後～

血縁者が遺伝子変異を持っている可能性と遺伝カウンセリングの提案

It is also possible that there is an identifiable mutation in your family in one of the genes tested that you did not inherit. Therefore, your maternal cousin with breast cancer in her early 40s could consider hereditary cancer genetic testing. If they live in the area, we would be happy to see them at the Stanford Cancer Genetics Clinic. If they live elsewhere, they can locate a genetics professional in their area by using the "Find a Genetic Counselor" tool on the following webpage: www.nsgc.org. Please update us should other relatives pursue testing, as their results have the potential to alter our interpretation of your negative test results and/or medical management considerations for you.

Your genetic testing was not able to identify an underlying hereditary cause to your personal and/or family history of cancer. There remains the possibility of a hereditary cause to the cancer in your family that we cannot determine at this time. You and your family members should continue to base medical management on personal and family history.

The future risk for cancer for you and your relatives must currently be estimated from empiric risk data. **Please refer to your consultation report for specific screening recommendations based on your**

検査の限界

Family Letter(3/4)
～家族性腫瘍 遺伝学的検査の結果開示後～

既往歴と家族歴に基づいた今後のがんの発症リスクと検診

personal and/or family history. Our cancer genetics physician is available to discuss recommendations if you wish to schedule an appointment. To briefly summarize:

- Based on your personal and family history, your future risk for a primary breast cancer is estimated to be 21% using the Tyrer-Cuzick model. Thus, you qualify for annual breast MRI screening in addition to annual mammogram.
- Depending on their personal risk factors and your personal and family history, your first degree female relatives (daughters and sisters) may have a 20% or greater lifetime breast cancer risk. This is within the range (>20- 25% lifetime risk) for which the American Cancer Society recommends annual breast MRI in addition to annual mammogram for breast cancer screening. Typically this screening is started by age 40 or 10 years prior to the earliest breast cancer diagnosis in the family, whichever is earlier. Your female relatives should discuss this increased level of breast screening with their physician. An appointment with our cancer genetics physicians is available should any of your female relatives be interested in a more detailed discussion.
- Based on your family history, colonoscopy screening every 5-10 year or per colonoscopy findings is appropriate for you and your siblings. You should discuss this screening with your physician.
- You do not have a family history of ovarian cancer and no gene mutations were detected in a hereditary ovarian cancer gene, thus we have no reason to suspect you are at increased risk for ovarian cancer over that of the general population (1 -2%). Screening and prevention strategies are not given for women at average risk.

Your children are not at-risk to carry a mutation in any of these genes from your genetic contribution, as they cannot carry a mutation for which you have tested negative. Therefore, genetic testing of these genes is not indicated for your children. If your children have a significant family history of cancer on their other parent's side of their family, then cancer genetic testing may be indicated for them. They may remain at elevated risk for cancer in adulthood based on their family history. This should be discussed with their doctor. In rare circumstances, children can be found to carry a gene mutation for which their parents have tested negative. This remains a theoretical risk.

次世代への遺伝

Family Letter(4/4)
～家族性腫瘍 遺伝学的検査の結果開示後～

DNAバンクについて

DNA banking is an option for every patient. The purpose is to preserve the option of additional genetic testing in the future, as genetic testing improves. DNA banking is not a test but rather a service provided by Prevention Genetics Lab (as well as other labs). The website and phone for Prevention Genetics are www.preventiongenetics.com; 715-387-0484.

Cancer genetics is a rapidly evolving field. New information regarding inherited cancer risks, genetic testing, prevention and treatment options will certainly change with time. Furthermore, the assessment we provided depends on the accuracy of the personal and family medical information you reported at the time of our consultation. If this information changes, it may affect your cancer risk and our recommendations. We invite all patients to re-contact our office every two to three years to discuss whether new developments might alter the medical management and/or additional genetic testing considerations outlined above. We ask patients to assume responsibility for future follow up once they have been discharged from our clinic. Please call us if you have additional questions or would like to schedule a follow up appointment. We can be reached at (650) 498-6000.

Sincerely,

[redacted] MS, CGC
Licensed Genetic Counselor
Cancer Genetics and Genomics

900 Blake Wilbur Drive
Stanford, CA 94305

[redacted]