A novel approach to lab-based clinical genetic counseling

KATE LYNCH, MS, LCGC
GENETIC COUNSELOR
INVITAE
Disclosure

I am an Invitae employee and stockholder.
Genetic testing has changed

- Decreased cost of genetic testing combined with improvements in technology have led to an unprecedented era of multi-gene panel testing.

- Demand for GC services is escalating without a parallel increase in the number of genetic counselors.

- To fill this void, some commercial laboratories employ genetic counselors to provide genetic counseling directly to patients undergoing testing in their lab.

- This raises potential for conflict of interest.

Conflict of interest

- **Realities**
  - Demand for genetic counseling cannot be met with the current number of GCs.
  - Specialized clinics are mainly in urban areas and academic centers, which limits access.
  - COI is an issue for all GCs, regardless of their employment setting.

- **Concerns**
  - Can laboratory GCs offer objective, unbiased information if employed by a company dependent upon test volume?
  - Might genetic counselors steer patients to proceed with testing not clinically indicated?
  - Might they be incentivized to recommend more testing than necessary?

- **Opportunities**
  - Use of lab-based GCs may allow patients access who would not otherwise have been able to receive services.

Recommendations/guidelines

- No published recommendations or professional guidelines exist to direct lab-based GCs providing clinical services.

- NSGC Task Force was created this year to explore COI among genetic counselors.

- Labs must develop internal policies.
• Invitae has made extensive efforts to demonstrate responsible and transparent genetic counseling services by defining a specific 5-point guideline for provision of patient care.
Invitae’s guideline for provision of care is based on five core principles

1. Abide by NSGC Code of Ethics

2. No commission; no quotas

3. Transparency with patients

4. Genetic counseling provided post-requisition

5. No additional cost for adding clinically appropriate genes
What does this look like in practice?
Pilot Study

- Three-way collaboration among Invitae, a large primary care practice, and a large private payer

- Primary care physicians were provided with referral criteria and a simple, streamlined process for referring patients for genetic testing and counseling.

- 129 referred patients underwent cancer genetic counseling during a 14-month period
Outcomes of genetic counseling

129 patients underwent genetic counseling

- 16% (21) test orders cancelled or altered
- 84% (108) test orders unchanged

- 57% (12) testing altered
- 43% (9) testing cancelled

- 33% (4) test orders decreased
- 66% (8) test orders increased
Outcomes of genetic counseling

129 patients underwent genetic counseling

- 16% (21) test orders cancelled or altered
- 57% (12) testing altered
- 33% (4) test orders decreased
- 43% (9) testing cancelled
- 66% (8) test orders increased
- 84% (108) test orders unchanged
Outcomes of genetic counseling

129 patients underwent genetic counseling

- 16% (21) test orders cancelled or altered
- 84% (108) test orders unchanged

- 57% (12) testing altered
- 43% (9) testing cancelled

- 33% (4) test orders decreased
- 66% (8) test orders increased
Outcomes of genetic counseling

- 129 patients underwent genetic counseling
  - 16% (21) test orders cancelled or altered
  - 84% (108) test orders unchanged
    - 57% (12) testing altered
    - 43% (9) testing cancelled
      - 33% (4) test orders decreased
      - 66% (8) test orders increased
Outcomes of genetic counseling

129 patients underwent genetic counseling

- 16% (21) test orders cancelled or altered
- 84% (108) test orders unchanged
- 57% (12) testing altered
- 43% (9) testing cancelled
- 33% (4) test orders decreased
- 66% (8) test orders increased

Increase: Due to identification of additional risk factors
Decrease: Due to previously identified mutation in a family member
Outcomes of genetic counseling

129 patients underwent genetic counseling

- 16% (21) test orders cancelled or altered
- 84% (108) test orders unchanged

**Increase:**
Due to identification of additional risk factors

**Decrease:**
Due to previously identified mutation in a family member

- 57% (12) testing altered
- 43% (9) testing cancelled
- 33% (4) test orders decreased
- 66% (8) test orders increased

1. Unaffected patient with a more appropriate affected family member to test
2. Patient deemed low risk after genetic counseling; declined testing
3. Inappropriate testing originally ordered
4. Affected family member already tested negative
Outcomes of genetic testing

Test results for 113 patients

- Positive: 11 (10%)
- VUS: 33 (29%)
- Negative: 69 (61%)
Conclusions

- These data provide an example of a principled application of lab-based genetic counseling services.

- A substantial number of patients had their genetic testing altered or cancelled after genetic counseling.

- A significant proportion of patients had test results that altered their clinical management.

- This model provides an example of how lab-based GC services can ensure the most appropriate test will be provided and improve access for optimal patient care.
Thank you!
kate.lynch@invitae.com


References


