Your abstract submission has been received

Click here **to print this page now.**

You have submitted the following abstract to the 2017 ASCO Annual Meeting (June 2-6, 2017). Receipt of this notice does not guarantee that your submission was complete, free of errors, or accepted for presentation. Abstract notifications will be sent to the First Author by April 3, 2017.

Germline mutations in cancer predisposition genes among patients with thyroid cancer.

Junne Kamihara, Holly LaDuca, Emily Dalton, Virginia Speare, Judy Ellen Garber, Mary Helen Black; Dana-Farber Cancer Institute, Boston, MA; Ambry Genetics, Aliso Viejo, CA

Abstract Text:

Background: Thyroid cancers are known component tumors of both well-described and emerging hereditary cancer syndromes. To assess the contribution of germline variants in thyroid cancer predisposition, we examined the prevalence of germline mutations among individuals with a history of thyroid cancer, compared to those with thyroid and breast cancer or breast cancer alone. **Methods:** Clinical histories and molecular results were reviewed for individuals with a history of thyroid and/or breast cancer, ascertained from a cohort of > 140,000 patients who underwent hereditary cancer multigene panel testing at a single commercial laboratory. Clinical history information was obtained from test requisition forms completed by ordering clinicians and from pedigrees/clinic notes, if provided. **Results:** Among 2,678 thyroid cancer patients, the majority were Caucasian (66.9%), female (92.3%), and/or had an additional cancer primary (71.9%), with nearly half reporting an additional breast cancer primary (49.1%). Among those with available pathology information, 4.1% had medullary thyroid cancer. The median (IQR) age at diagnosis was 38 (26,48) years, and while 94.1% had a family history of cancer, 78.8% had at least one affected 1st degree relative. Overall, 11.1% were identified as mutation carriers, defined as ≥1 pathogenic or likely pathogenic variant. Among those with thyroid cancer alone, 9.7% had a mutation, similar to those with breast cancer alone (9.7%) and those with breast and thyroid cancer only (10.5%). Genes most frequently mutated in the thyroid only group included *CHEK2* (3.1%), *MUTYH* (monoallelic) (2.4%), *APC* (2.0%), *ATM* (1.6%), and *PALB2* (1.2%). *CHEK2* was the most frequently mutated gene observed in all groups, with a higher frequency seen among those with thyroid and breast cancer (5.5%) compared to breast cancer (2.5%) or thyroid cancer (3.1%) alone ($p < 0.001$). **Conclusions:** A high rate of germline mutations is observed among individuals with thyroid cancer presenting for clinical genetic testing, even in the absence of other primary cancer diagnoses. Thyroid cancer may be an under-recognized component tumor of hereditary cancer predisposition syndromes suggesting the need for further investigation.

Title:
Germline mutations in cancer predisposition genes among patients with thyroid cancer.

Submitter's E-mail Address:
junne_kamihara@dfci.harvard.edu

Is this a late-breaking data submission?
No

Is this abstract a clinical trial?
No

Would like to be considered for a Merit Award:
No

Have the data in this abstract been presented at another major medical meeting?
No

Has this research been submitted for publication in a medical journal?
No

Type of Research:
Exploratory Analysis
Research Category:
Translational
Continued Trial Accrual:
No
Received Grant funding:
Yes - Young Investigator Award (YIA), 2014
Relevant to geriatric oncology:
No
Sponsor:
Junne Kamihara, MD, PhD

First Author

Presenting Author
Corresponding Author
Junne Kamihara, MD, PhD
Dana-Farber Cancer Institute
450 Brookline Ave
Boston, MA 2215
Email: junne.kamihara@childrens.harvard.edu
Alternate Email: junne_kamihara@dfci.harvard.edu

Click to view Conflict of Interest Disclosure

Second Author

Holly LaDuca, MS, CGC
Ambry Genetics
Aliso Viejo, CA

Email: HLADUCA@AMBRYGEN.COM

Click to view Conflict of Interest Disclosure

Third Author

Emily Dalton, MS
Ambry Genetics
Aliso Viejo, CA
Email: edalton@ambrygen.com

Click to view Conflict of Interest Disclosure

Fourth Author

Virginia Speare, PhD
Ambry Genetics
15 Argonaut
Aliso Viejo, CA 92656
Email: vspeare@ambrygen.com

Click to view Conflict of Interest Disclosure

Fifth Author

Judy Ellen Garber, MD, MPH
Dana-Farber Cancer Institute
Boston, MA 02215
Phone Number: 617-632-2282
Alternate Phone: 617-763-8821
Email: judy_garber@dfci.harvard.edu
Alternate Email: audrey_kalisz@dfci.harvard.edu

Click to view Conflict of Interest Disclosure

Sixth Author

Mary Helen Black
Ambry Genetics
Aliso Viejo, CA
Email: mblack@ambrygen.com

Click to view Conflict of Interest Disclosure

If necessary, you can make changes to your abstract between now and the deadline of Tuesday, February 7, 2017

- To access your submission in the future, use the direct link to your abstract submission from one of the automatic confirmation emails that were sent to you during the submission.
- *Or* point your browser to /asco/reminder.cgi to have that URL mailed to you again. Your username/password are 190158/124225.

Any changes that you make will be reflected instantly in what is seen by the reviewers. You DO NOT need to go through all of the submission steps in order to change one thing. If you want to change the title, for example, just click "Title" in the abstract control panel and submit the new title.

When you have completed your submission, you may close this browser window.

Tell us what you think of the abstract submission process

Home Page