Abstract

Angiosarcoma (AS) is a very rare soft tissue carcinoma, with an incidence of 0.33 cases/100,000 in 2010. There are very limited insights into the underlying biology of AS and there is no standard of care for treatment. To address this need, we launched a genome-wide clinical genomics project to build knowledge on the biology driving this rare tumor and improve personal care. Patient specimens were obtained from multiple institutions and the research team is conducting whole-genome sequencing and whole-exome sequencing on all patient samples. Over 500 patients have been enrolled, and analyses will be conducted at scale to build the largest AS dataset to date. These analyses will be shared with the scientific community through the LSRCC data portal (cBioPortal.org).

Challenges of Studying Rare Patient Tumor Samples

- Only 5% of U.S. cancer patients are diagnosed with AS.
- 87% of U.S. patients are treated at a single institution.
- Genetic testing is only performed in ~7% of patients.

Participating Directly with Patients

- ASDC is an open-access program that provides a free opportunity for patients to participate directly in cancer research through the ASDC project.
- ASDC is committed to genotyping saliva samples from patients with AS.

Participation in The Angiosarcoma Project to Date

<table>
<thead>
<tr>
<th>Date</th>
<th>Participants</th>
<th>DNA Sequencing on Blood</th>
<th>DNA Sequencing on Saliva</th>
</tr>
</thead>
<tbody>
<tr>
<td>1/1/2020</td>
<td>250</td>
<td>200</td>
<td>0</td>
</tr>
<tr>
<td>3/1/2020</td>
<td>400</td>
<td>300</td>
<td>100</td>
</tr>
<tr>
<td>6/1/2020</td>
<td>500</td>
<td>400</td>
<td>100</td>
</tr>
<tr>
<td>9/1/2020</td>
<td>600</td>
<td>500</td>
<td>100</td>
</tr>
<tr>
<td>12/1/2020</td>
<td>700</td>
<td>600</td>
<td>100</td>
</tr>
</tbody>
</table>

Preliminary Sequencing Results

<table>
<thead>
<tr>
<th>Probes</th>
<th>Number of Samples</th>
<th>Proportion</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tumor</td>
<td>100</td>
<td>100%</td>
</tr>
<tr>
<td>Blood</td>
<td>100</td>
<td>100%</td>
</tr>
</tbody>
</table>

Over 700 women and men with angiosarcoma from 73 sites have joined the ASDC project since our launch. Many patients have made contributions to science as a way of sharing the experience of a loved one who died from angiosarcoma.

Loved One Survey Data

<table>
<thead>
<tr>
<th>Question</th>
<th>Response</th>
</tr>
</thead>
<tbody>
<tr>
<td>What therapies did you and your loved one try?</td>
<td>Radiation therapy, surgery, and chemotherapy</td>
</tr>
<tr>
<td>What were the biggest challenges that you faced?</td>
<td>Financial burden, lack of information, and logistical barriers</td>
</tr>
<tr>
<td>What do you think is the future of angiosarcoma research?</td>
<td>Potentially curative treatments are on the horizon</td>
</tr>
</tbody>
</table>

Acknowledgements

We are grateful to all the patients, their advocates, and the healthcare providers who have participated and have contributed to this project. We thank our partners, including the LSRCC, the Broad Institute, and the National Cancer Institute. We also gratefully acknowledge the contributions of the Advocacy Partners and the many others who have supported this project. We thank the patients and families who have shared their stories with us for their guidance and support.

The Angiosarcoma Project: Generating the genomic landscape of an exceedingly rare cancer through a nationwide patient-driven initiative


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Supporting Information

- PDF: The Angiosarcoma Project: Generating the genomic landscape of an exceedingly rare cancer through a nationwide patient-driven initiative
- HTML: The Angiosarcoma Project: Generating the genomic landscape of an exceedingly rare cancer through a nationwide patient-driven initiative
- JSON: The Angiosarcoma Project: Generating the genomic landscape of an exceedingly rare cancer through a nationwide patient-driven initiative
- CSV: The Angiosarcoma Project: Generating the genomic landscape of an exceedingly rare cancer through a nationwide patient-driven initiative
- XML: The Angiosarcoma Project: Generating the genomic landscape of an exceedingly rare cancer through a nationwide patient-driven initiative

The Patient Voice

"I want my daughter to be the one to decide whether she wants to participate in this project. I want her to have the choice."

Lauren Ryan

Summary and Conclusions

- This project is the largest ever patient-driven genomics project for angiosarcoma. It is unique in that patients and their families are the driving force behind this project.
- The project is supported by a network of partners and organizations, including the LSRCC, the LSRCC, and the National Cancer Institute.
- The project is open to all patients with angiosarcoma, and their families are encouraged to participate.

Angiosarcoma Diagnosis Location (N=294)

- Sites of spread: Scalp, 21%; Abdominal, 17%; Limb, 15%; Other, 5%
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