



EHE BIOBANK | PATIENTS POWERING RARE CANCER MODEL DEVELOPMENT & TRANSLATIONAL RESEARCH

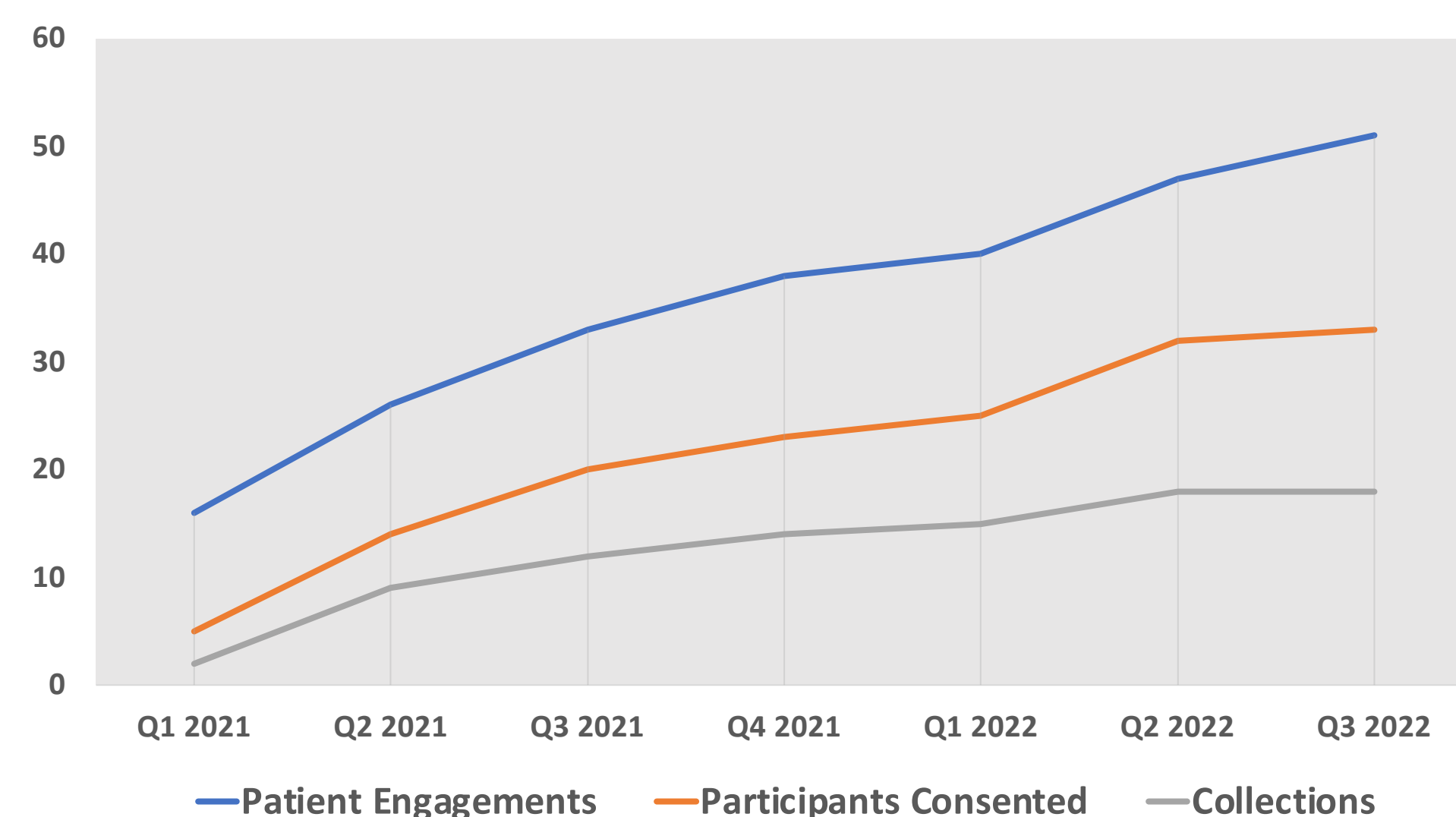
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OBJECTIVES

Epithelioid Hemangioendothelioma (EHE) is an ultra-rare vascular sarcoma with a prevalence of less than 1 per million people.¹ The rarity of this disease, like many rare cancers, presents significant challenges in basic and translational research. In addition to the rare incidence of EHE, researchers studying EHE may not be affiliated with a center treating EHE patients, limiting the opportunity to collect EHE tumor tissue and other biospecimens. EHE biospecimens that are obtained during procedures or surgeries are most often retained at patients' treating centers and are not readily available to researchers. These challenges inhibit EHE model development, as well as basic and translational research due to insufficient availability of EHE biospecimens.

There are relatively few scientists working directly with EHE specimens and today, there are no human EHE cell lines in existence. Recently, the publication of the first EHE PDX (patient derived xenograft) model has been developed and is available²; additionally, one EHE GEM model (genetically engineered mouse) was developed in 2021³. To address the unmet need of biospecimens required to advance EHE research, The EHE Foundation initiated the EHE Biobank: a collection of biospecimens and clinical data to facilitate research. The EHE Biobank is a patient-led initiative aiming to centralize EHE biospecimens, making specimens readily available for ethical research while actively seeking research partners to develop additional EHE models. The Biobank collects fresh EHE tumor tissue from prospective surgeries as well as archived EHE tumor tissue. Additionally, the Biobank collects serosal effusions, blood, and saliva.

Patient Engagement & Specimen Collection



Specimen Collection & Distribution	No.
Participants with Data	33
Participants Donated Specimens	17
Tissue Specimens Collected	12
Fluid Specimens Collected	6
Normal-Adjacent Tissue Collected	1
Matched Blood Samples	4
Specimens Distributed to Researchers	15

METHODS

The EHE Foundation engaged expert pathologists and clinicians in the sarcoma medical community, disease-expert patient advocates, EHE researchers, and other rare cancer advocacy organizations to develop a centralized collection of EHE biospecimens. An expert scientist in the field of biobanking, specifically for rare cancers, was enlisted to serve as the EHE Biobank Principal Investigator and Coordinator. A working team was formed to develop the Biobank protocol, Informed Consent templates, and work processes.

The Foundation took great care in order to identify the institution best suited to serve as the central repository for this ultra-rare cancer. We anticipated fewer than 50 samples per year and assumed close attention would be required during each specimen collection in order to maximize the utilization of each specimen and expedite distribution of specimens to researchers. The Cleveland Clinic BioRepository was selected for its highly skilled team, the capability to provide tailored processes and access to an expert pathologist for a confirmatory assessment of each specimen to verify the EHE tumor-defining gene fusion.

The protocol and supporting documents were reviewed and approved by North Star Review Board, a central Independent Review Board. The protocol was designed to collect biospecimens from participants in the United States only. Specimen collection began in February 2021.

The EHE Foundation is using several mechanisms for patient engagement including its website, www.fightehe.org, newsletters, direct emails, in social channels and in private online patient communities.

A process for participant engagement was developed to ensure consistency in communications, effective reporting and follow-up from the earliest points of contact with each interested participant. The Biobank Coordinator facilitates the Informed Consent process virtually with each prospective Biobank participant. Informed Consent is administered electronically.

RESULTS

The EHE patient community is highly engaged with The EHE Foundation, readily utilizing The Foundation's Patient Services team as a resource. Leveraging patient engagement among an active private network of over 2,000 EHE patients and advocates, The EHE Foundation actively promotes the Biobank via social media, patient conferences, events, by email and newsletters, and by communicating with clinicians treating EHE patients.

At this time 51 patients have engaged with the EHE Biobank Coordinator, 33 participants have provided Informed Consent, 18 EHE biospecimens have been collected. Of the specimens collected 15 have been distributed to researchers.

Additionally, engagement among medical and scientific stakeholders in the EHE community has grown significantly during the past 1.5 years. The Foundation has enlisted partner researchers to attempt to develop EHE cell lines using fresh tissue distributed from the EHE Biobank, and while no cell lines have yet been confirmed, we are optimistic for success in the near term. The Foundation is also assessing what additional data we would like to generate on collected biospecimens that could provide information to the scientific community and drug developers.

Key learnings emerged early-on after the project was initiated, revealing the importance of confirming EHE in each sample collected. Because EHE, like other sarcoma sub-types, is commonly misdiagnosed, The EHE Foundation recognized the need to return any findings indicating that a biospecimen was not confirmed as EHE. Thus, the Biobank protocol was amended to permit the return of any such finding to a participant's care provider engaged during the collection process with a goal to better inform participant medical care.

CONCLUSIONS

Patient advocates and people living with EHE have the ability to significantly advance rare cancer research and are uniquely positioned to accelerate EHE model development by donating human specimens. Patients' care teams are integral to this process and are essential in helping advocates educate patients about the importance of contributing to this non-interventional research. In summary, a collaborative approach to engage participants, centralize collected specimens, and actively distribute EHE biospecimens is essential to advancing translational research.

Ongoing active engagement of the EHE community, including advocates, patients, clinicians, and researchers is vital to increase awareness of the importance of specimen donation and its impact on EHE research.

REFERENCES

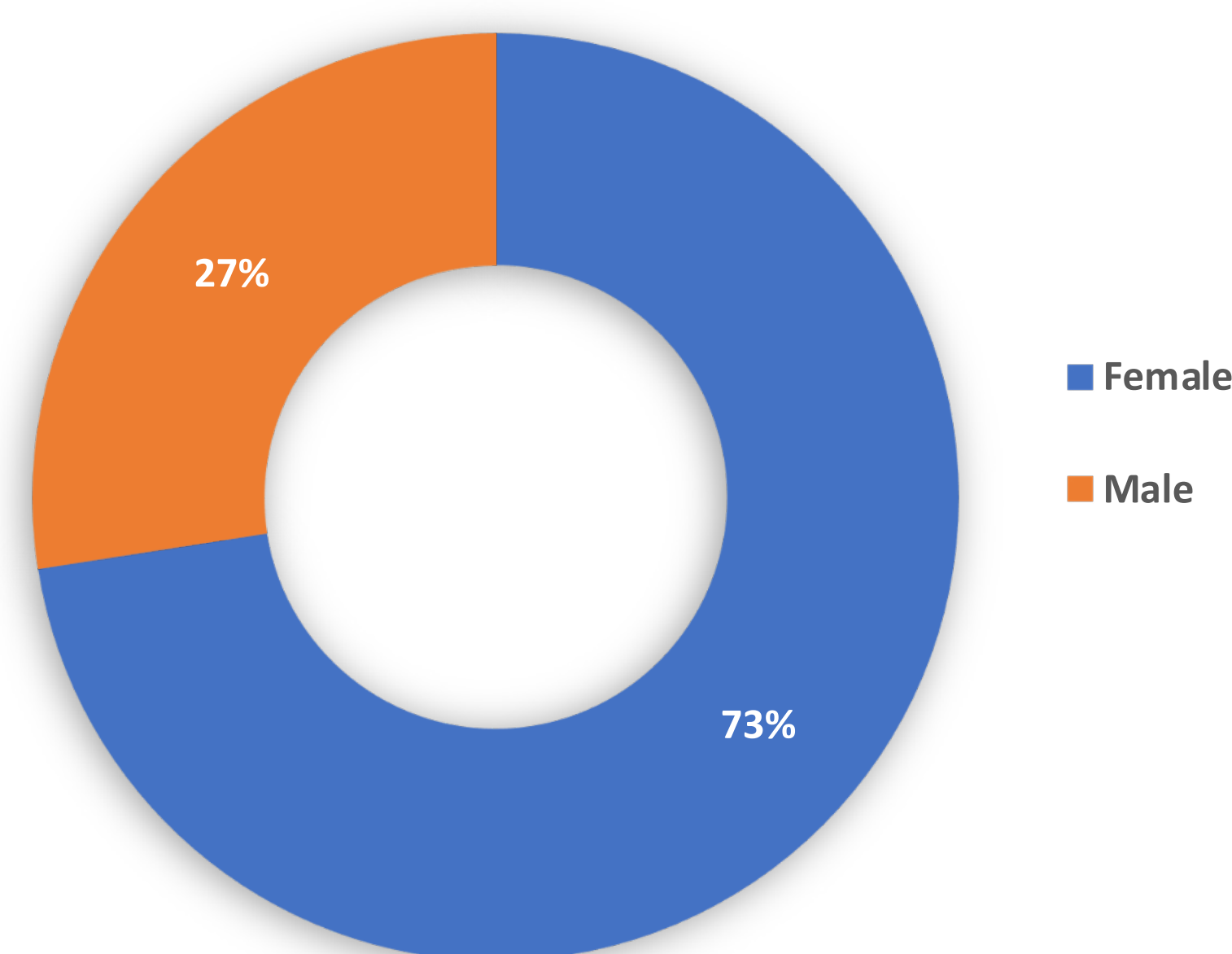
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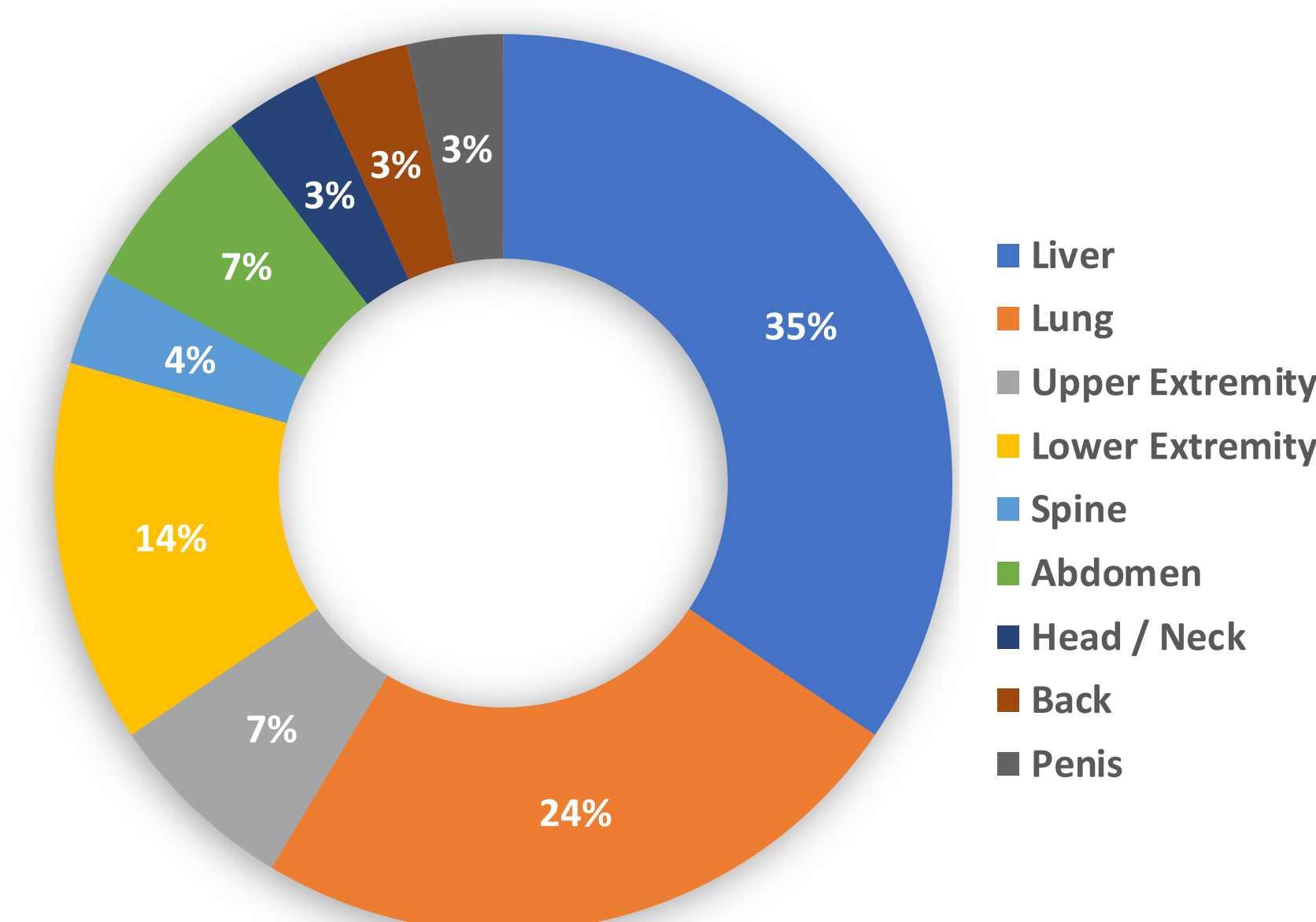
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The EHE Foundation is grateful to patients who have donated specimens to the EHE Biobank. These patients have given an invaluable contribution toward the advancement of EHE research, and with their gift they give hope for a world where EHE is effectively treated and cured.

Engagement by Gender



Original Tumor Location
(of Consented Participants)



Specimen Collection by Disease State

