Good, Better, Best: Envisioning the Future

Connie Lee, President and CEO, Angioma Alliance
Our Mission

To inform, support, and empower those affected by cavernous angioma and drive research for better treatments and a cure.
The Full Picture

- Diagnosis
- Tracking/Predicting
- Lesion Treatment Options
- Multi-Systemic Impacts

- Recovery/Restoration
- Comprehensive Clinical Care
- Supportive Community
2006: Where We Were

- Inconsistent diagnosis
- Few natural history studies
- Surgery/radiosurgery were the only interventions
- What multi-systemic effects?
- Basic rehab
- What prevention?
- A few knowledgeable surgeons and fewer neurologists
- Loose patient community
More questions than answers
2018: Where We Are

- Diagnosis - SWI MRI; genetic testing (Angioma Alliance)
- Course - Natural history data regarding hemorrhage, incidental lesions, genetics, proliferation.
- Treatment - Surgery, Minimally invasive surgery, Vitamin D
- Identifying multi-systemic issues and causes/consequences
2018: Where We Are

- **Rehab** - More informed PT, OT, surgery, orthotics, robotics
- **Prevention** - Option of IVF with PGD
- **Care** - 7+ US Clinical Centers recognized for providing expert multi-disciplinary care
- **Community** - We can find each other, offer support, participate in advocacy/research/raising funds.
How Did We Get Here?

- Genes identified/testing
- Scientific Meeting to advance collaborations
- DNA/Tissue Bank
- Legislation and advocacy
- SAIVMS, BVMC Project and other natural history studies
- Centers of Excellence
- Community building
You're Awesome!
Accurate diagnosis - urine biomarkers (Boston Children’s), blood biomarkers, genetic testing covered by insurance, shorter MRI

Accurate course prediction - blood and imaging biomarkers (U of Chicago)

Medications and non-invasive treatments to stabilize vessels, reduce hemorrhage risk, reduce proliferation, reduce post-surgical recurrence

Reduced multi-systemic impact of hereditary disease

2024: Real Treatment Options!
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- Restoration of lost function - stem cell therapy trials? (Stanford)
- Prevention using microbiome change via individual diet change (CCM diet)/transplants, cheaper IVF with PGD
- Many Clinical Centers providing expert care around the world
- A community that is organized and mobilized for research participation, fund and awareness raising, and outreach to clinicians/patients.
Path to the Future
How Do We Get There?

- Participate and/or support clinical trials and other clinical research: join patient registry, get testing, educate peers.
- Support basic/translational research: scientific meeting, advocacy, grants?
- Form many Community Alliances for outreach to medical community, etc.
- Increase international patient participation in research.
Working Together Will Speed Progress!

Oceania: 38,000,000
South America: 422,000,000
North America: 579,000,000
Europe: 741,000,000
Africa: 1,216,000,000
Asia: 4,463,000,000
2030: The Final Destination

- Fast, cheap, and accurate diagnosis - urine biomarkers, $50
genetic testing, $200 10-minute MRI
- Fast, cheap, 100% accurate course prediction -
  blood/imaging biomarkers, something else?
- Obliterate lesions with no recurrence or side effects using a
  low-cost one-time medication or non-invasive treatment.
2030: The Final Destination

- Eliminate multi-systemic impact of hereditary disease
- Cheap restoration of lost function as well as development of functions that didn’t have a chance in those affected as young children.
- Societal microbiome changes.
- Gene therapy for prevention of generational transmission.
2030: The Final Destination

- Informed medical providers everywhere, even primary care.
- We will be known in all medical settings and among the general public.
- The cure, prevention, and restorative treatments will be readily available.
How Do We Get To a Cure?

- Do all the same things we do for better treatments! Organize, mobilize, participate!
- Create a tandem, alternate route that supplements the pharmaceutical industry - just in case.
How Do We Get To a Cure?

- Partner with other disease groups internationally whose illnesses may be amenable to same cure.
- Partner with governments, industries outside of pharma, and foundations.
- Fundraise to fill gaps in the development of cures.
Take-Aways

- We are in a **new era** for cavernous angioma.
- This era will include better prediction, non-surgical treatments, informed care, and mobilized, supported patients.
- We are moving forward because of YOU and those around you!