

# Clinical Atlas of Variant Effects Workshop

Developing recommendations to produce a maximally useful Clinical Atlas of Variant Effects by 2030.

## Location and details

July 23rd, 2024

8am-9am Arrival, breakfast, and funders meeting

9am-5pm Workshop (includes lunch)

7pm-9pm Dinner

The Oaklander Hotel, The Cathedral Room  
5130 Bigelow Boulevard, Pittsburgh, Pennsylvania 15213

## Meeting materials

- [Event Website](#)
- [Participant Bios](#)
- [Short explainer](#)
- [Straw Proposals](#)
- [Slide deck for report outs](#)

## Agenda overview

- I. Welcome and charge for the day
- II. Setting the stage and straw proposal introduction
- III. Deep discussion of straw proposals - five year goals and scope
- IV. Strengths, weaknesses and opportunities based on where we are now
- V. Roadmap to 2030 - developing a tech, infrastructure, and coordination strategy
- VI. Synthesis, prioritization and action Items

**Clinical Atlas of Variant Effects Workshop**  
**Tuesday, July 23rd, 2024**  
**8:00am-5:00pm EDT**

*Location: All events in The Cathedral Room | Oaklander Hotel unless otherwise noted*

Start (EDT)	End (EDT)	Agenda item <i>Speaker(s)</i>
8:00am	9:00am	<b>Arrival, breakfast and coffee</b>
		<b>Funders breakfast</b> ( <i>The George Boardroom</i> )
9:00am	9:05am	<b>Welcome</b> <i>Frederick 'Fritz' Roth, University of Pittsburgh, USA</i>
9:05am	9:35am	<p><b>Charge for the day</b> (5 minutes)  <i>Jonah Cool, Chan Zuckerberg Initiative, USA</i>  <i>Carolyn Hutter, National Human Genome Research Institute, USA</i></p> <p><b>Technology</b> (5 minutes)  <i>Douglas Fowler, University of Washington, USA</i></p> <p><b>Infrastructure and community</b> (5 minutes)  <i>Frederick 'Fritz' Roth, University of Pittsburgh, USA</i></p> <p><b>Clinical translation</b> (5 minutes)  <i>Heidi Rehm, Broad Institute/Mass General Hospital, USA</i></p> <p><b>Data production</b> (5 minutes)            With a focus on Strengths and Weaknesses and Opportunities  <i>Matthew Hurles, Director Wellcome Sanger Institute, Hinxton, UK</i></p> <p><b>Session Goals:</b></p> <ul style="list-style-type: none"> <li>&gt; Introduce goals for the day</li> <li>&gt; Level setting/background</li> </ul>
9:35am	9:50am	<b>AVE 2030 straw proposals summary</b> <i>Douglas Fowler, Professor, University of Washington, USA</i> <i>Matthew Hurles, Director Wellcome Sanger Institute, Hinxton, UK</i>
9:50am	10:05am	<b>Whole-group discussion of straw proposals</b> <i>Moderator(s): Jonah Cool</i>
10:05am	10:35am	<b>Breakout discussion of straw proposal goals and scope</b> <i>Moderator(s): Fritz Roth</i> <b>Goal:</b> Identify high-level goal(s) for an atlas in a five year timeframe

		<p><b>Task:</b> Join one of the 6 breakout groups listed below, aiming for ~6 individuals per group. Discuss goals and scope for an atlas, with a five year timeframe in mind. Your group might refine straw proposal goals, combine them or advance entirely different ones. Use the documents below to take notes or develop ideas.</p> <p>Breakout 1: (<a href="#">google doc link</a>)          Breakout 2: (<a href="#">google doc link</a>)          Breakout 3: (<a href="#">google doc link</a>)          Breakout 4: (<a href="#">google doc link</a>)          Breakout 5: (<a href="#">google doc link</a>)          Breakout 6: (<a href="#">google doc link</a>)</p>
10:35am	10:45am	<p><b>Breakout group wrap up and synthesis</b></p> <p><b>Task:</b> Capture your breakout discussion and conclusions in the slides linked below. We suggest allotting some time for independent reflection and note-writing as well as for breakout group discussion. Identify a reporter to present a two minute summary using the slide(s) provided.</p> <p><a href="#">Slide deck for report outs</a></p>
10:45am	11:15am	<b>Break</b>
11:15am	11:45am	<p><b>Breakout report out and discussion</b>  <i>Moderator for this session: Fritz Roth</i></p> <ul style="list-style-type: none"> <li>➤ Breakout groups report back (two minutes per group)</li> <li>➤ Whole group discussion of straw proposal goals and scope</li> </ul>
11:45am	11:50am	<b>Group photo</b>
11:50am	12:50pm	<b>Lunch</b>
12:50pm	2:00pm	<p><b>Where we are now: strengths, weaknesses and opportunities</b>  <i>Moderator(s) for this session: Carolyn Hutter and Lara Muffley</i></p> <p><b>Goals:</b></p> <ul style="list-style-type: none"> <li>➤ Identify what is working well and begin to identify how we can build on momentum</li> <li>➤ Identify what is working poorly or not working at all, and identify how we can gain momentum or set these items aside for later</li> </ul> <p><b>Task:</b></p> <ul style="list-style-type: none"> <li>➤ Brainstorm strengths, weaknesses and opportunities (one station for each) of current efforts towards building a Clinical Atlas of Variant Effects. Give considerations to the following categories (~10 min).</li> </ul>

		<ul style="list-style-type: none"> <li>○ Technology</li> <li>○ Infrastructure</li> <li>○ Clinical translation</li> <li>○ Data production</li> <li>○ Other</li> </ul> <ul style="list-style-type: none"> <li>➤ Rotate through the three stations to share/discuss/prioritize (~30 min)</li> <li>➤ Summarize strengths, weaknesses and opportunities (each group reporting back; ~20 min)</li> </ul>
2:00pm	2:20pm	<b>Break</b>
2:20 pm	3:40pm	<p><b>Defining a roadmap to 2030</b>  <i>Moderator for this session: Heidi Rehm</i></p> <p><b>Session Goals:</b></p> <ul style="list-style-type: none"> <li>➤ Identify major activities needed to achieve 2030 goals</li> <li>➤ Develop plans to facilitate identified activities</li> <li>➤ Consider who should do the work and how they should be incentivized/motivated</li> <li>➤ Identify resources that are needed from other groups/consortia</li> <li>➤ Discuss coordination between labs and consortia</li> </ul> <p><b>Task:</b></p> <ul style="list-style-type: none"> <li>➤ Join your pre-assigned breakout group, which is noted on the back of your name-badge</li> <li>➤ Contemplate the goals of this session and use the question prompts to lead discussions.</li> </ul> <p>Use the documents below for questions to consider and to take notes:</p> <p><a href="#">Clinical translation (Breakout group A)</a></p> <p><a href="#">Clinical translation (Breakout group B)</a></p> <p><a href="#">Data production (Breakout group A)</a></p> <p><a href="#">Data production (Breakout group B)</a></p> <p><a href="#">Infrastructure, standards &amp; community (Breakout group A)</a></p> <p><a href="#">Infrastructure, standards &amp; community (Breakout group B)</a></p> <p><a href="#">Technologies and tools - What is missing? (Breakout group A)</a></p> <p><a href="#">Technologies and tools - What is missing? (Breakout group B)</a></p>
3:40pm	3:50pm	<p><b>Breakout group wrap up and synthesis</b></p> <p><b>Task:</b> Capture your breakout discussion and conclusions in the slides linked below. We suggest allotting some time for independent reflection and note-writing as well as for breakout group discussion. Identify a reporter to present a two minute summary using the slide(s) provided.</p> <p><a href="#">Slide deck for report outs</a></p>

3:50pm	4:20pm	<p><b>Breakout report out and discussion</b>  <i>Moderator for this session: Heidi Rehm</i></p> <ul style="list-style-type: none"> <li>➤ Synthesis session (each group reporting back)</li> <li>➤ Facilitated discussion of the Roadmap to a Clinical Atlas of Variant Effects by 2030</li> </ul>
4:20pm	4:35pm	<b>Break</b>
4:35pm	5:05pm	<p><b>Recap and synthesis session</b>  <i>Douglas Fowler, Professor, University of Washington, USA</i>  <i>Matthew Hurles, Director Wellcome Sanger Institute, Hinxton, UK</i></p> <p><b>Session Goals:</b></p> <ul style="list-style-type: none"> <li>➤ Develop a coherent set of recommendations for international coordination to produce a maximally useful Atlas.</li> <li>➤ Generation of an improved proposal</li> </ul>
7:00pm	9:00pm	<b>Dinner</b> (Lucca Ristorante, 317 South Craig Street)