

A HISTORICAL REVIEW OF STRUCTURAL VARIANT DISCOVERY

Evan Eichler played a significant role in advancing the understanding of structural variants during the sequencing era. However, the exploration of structural variant discovery predates this era, commencing with cytogenetics.

In a recent paper (1), Eichler conducts a retrospective analysis of this narrative, emphasizing the current breakthrough achieved through the use of state-of-the-art long-read sequencing. He highlights the fact that we are now on the brink of achieving comprehensive variant detection across all forms of genetic variation using a single assay.

Some of these steps have been personally illustrated by Eichler during our past ECA conferences.

1. [https://www.cell.com/cell/fulltext/S0092-8674\(24\)00004-7?returnURL=https%3A%2F%2Flinkinghub.elsevier.com%2Fretrieve%2Fpii%2FS0092867424000047%3Fshowall%3Dtrue](https://www.cell.com/cell/fulltext/S0092-8674(24)00004-7?returnURL=https%3A%2F%2Flinkinghub.elsevier.com%2Fretrieve%2Fpii%2FS0092867424000047%3Fshowall%3Dtrue)