type of variant tested

A

2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
n = 2	n = 3	n = 4	n = 8	n = 13	n = 13	n = 18	n = 17	n = 20	n = 17

type of variant tested
- SNVs
- CNVs
- SNVs + CNVs

sequencing approach

B

2013	2014	2015	2016	2017	2018	2019	2020	2021
n = 1	n = 4	n = 3	n = 7	n = 12	n = 13	n = 12	n = 13	n = 14

sequencing approach
- enrichment
- exome-based

CNV approach

C

2012	2014	2015	2016	2017	2018	2019	2020	2021
n = 1	n = 4	n = 3	n = 7	n = 12	n = 13	n = 12	n = 13	n = 14

CNV approach
- single gene (e.g. MLPA)
- several genes (2-10)
- genome/exome wide (array)
- multiple genes (<100)
- multiple genes (>100)
- genome/exome wide (WES/WGS)
- unclear

ACMG classification

D

2012	2013	2014	2015	2016	2017	2018	2019	2020	2021
n = 2	n = 3	n = 4	n = 8	n = 13	n = 13	n = 18	n = 17	n = 20	n = 17

ACMG classification
- No
- Yes
- Yes + other