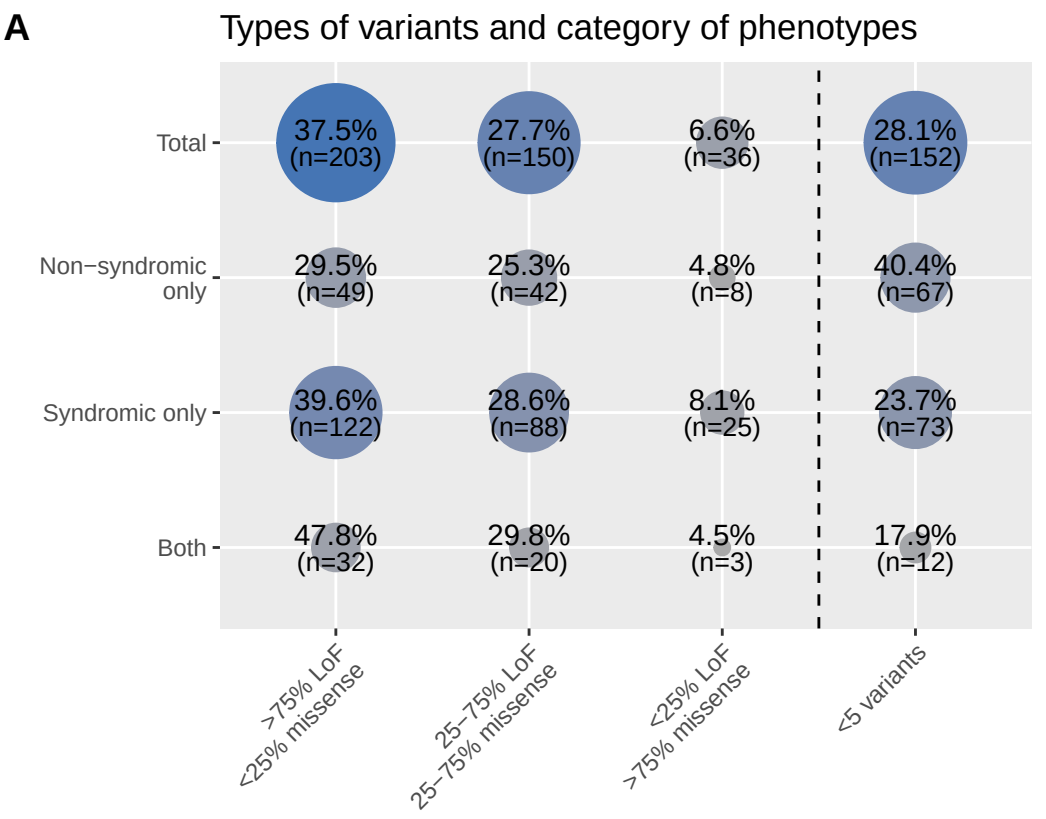
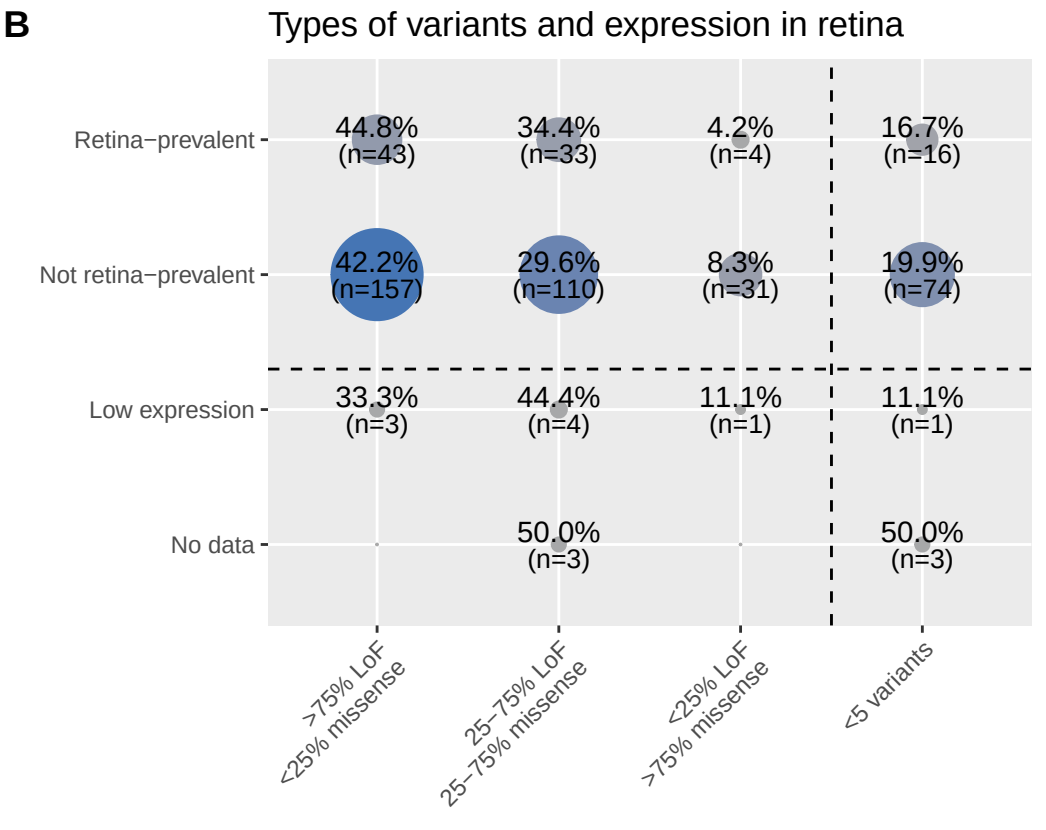

A
Types of variants and category of phenotypes
Total
Non−syndromic only
Syndromic only
Both
37.5% (n=203)
27.7% (n=150)
6.6% (n=36)
28.1% (n=152)
29.5% (n=49)
25.3% (n=42)
4.8% (n=8)
40.4% (n=67)
39.6% (n=122)
28.6% (n=88)
8.1% (n=25)
23.7% (n=73)
47.8% (n=32)
29.8% (n=20)
4.5% (n=3)
17.9% (n=12)
>75% LoF <25% missense
25−75% LoF 25−75% missense
<25% LoF >75% missense
<5 variants


B
Types of variants and expression in retina
Retina−prevalent
Not retina−prevalent
Low expression
No data
44.8% (n=43)
34.4% (n=33)
4.2% (n=4)
16.7% (n=16)
42.2% (n=157)
29.6% (n=110)
8.3% (n=31)
19.9% (n=74)
33.3% (n=3)
44.4% (n=4)
11.1% (n=1)
11.1% (n=1)
50.0% (n=3)
50.0% (n=3)
>75% LoF <25% missense
25−75% LoF 25−75% missense
<25% LoF >75% missense
<5 variants