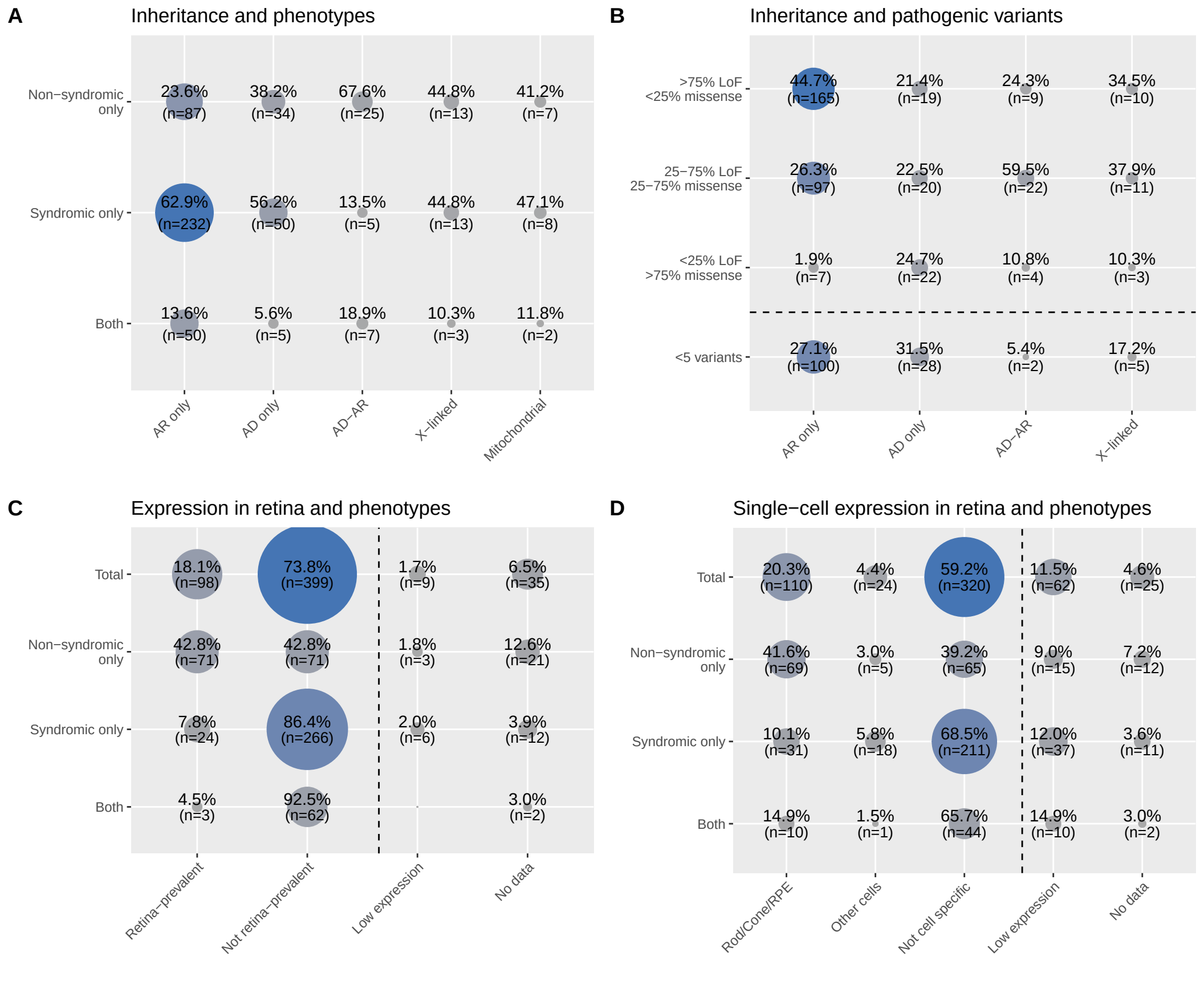
A
Inheritance and phenotypes
Non−syndromic only
Syndromic only
Both
23.6% (n=87)
38.2% (n=34)
67.6% (n=25)
44.8% (n=13)
41.2% (n=7)
62.9% (n=232)
56.2% (n=50)
13.5% (n=5)
44.8% (n=13)
47.1% (n=8)
13.6% (n=50)
5.6% (n=5)
18.9% (n=7)
10.3% (n=3)
11.8% (n=2)
AR only
AD only
AD−AR
X−linked
Mitochondrial
B
Inheritance and pathogenic variants
>75% LoF <25% missense
25−75% LoF 25−75% missense
<25% LoF >75% missense
<5 variants
44.7% (n=165)
21.4% (n=19)
24.3% (n=9)
34.5% (n=10)
26.3% (n=97)
22.5% (n=20)
59.5% (n=22)
37.9% (n=11)
1.9% (n=7)
24.7% (n=22)
10.8% (n=4)
10.3% (n=3)
27.1% (n=100)
31.5% (n=28)
5.4% (n=2)
17.2% (n=5)
AR only
AD only
AD−AR
X−linked
C
Expression in retina and phenotypes
Total
Non−syndromic only
Syndromic only
Both
18.1% (n=98)
73.8% (n=399)
1.7% (n=9)
6.5% (n=35)
42.8% (n=71)
42.8% (n=71)
1.8% (n=3)
12.6% (n=21)
7.8% (n=24)
86.4% (n=266)
2.0% (n=6)
3.9% (n=12)
4.5% (n=3)
92.5% (n=62)
3.0% (n=2)
Retina−prevalent
Not retina−prevalent
Low expression
No data
D
Single−cell expression in retina and phenotypes
Total
Non−syndromic only
Syndromic only
Both
20.3% (n=110)
4.4% (n=24)
59.2% (n=320)
11.5% (n=62)
4.6% (n=25)
41.6% (n=69)
3.0% (n=5)
39.2% (n=65)
9.0% (n=15)
7.2% (n=12)
10.1% (n=31)
5.8% (n=18)
68.5% (n=211)
12.0% (n=37)
3.6% (n=11)
14.9% (n=10)
1.5% (n=1)
65.7% (n=44)
14.9% (n=10)
3.0% (n=2)
Rod/Cone/RPE
Other cells
Not cell specific
Low expression
No data