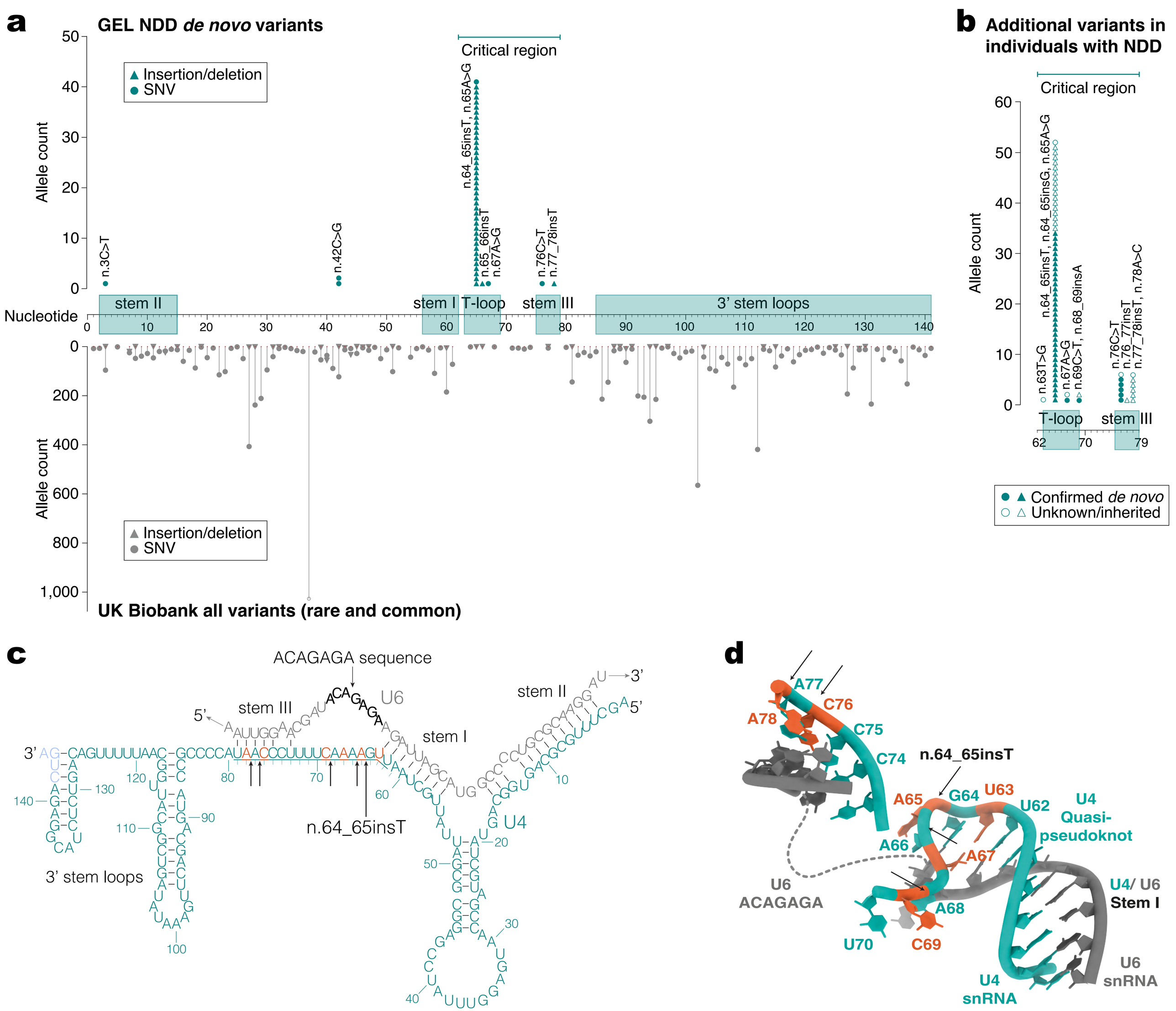
a
GEL NDD de novo variants
Allele count
Insertion/deletion
SNV
Critical region
n.3C>T
n.42C>G
n.64_65insT, n.65A>G
n.65_66insT
n.67A>G
n.76C>T
n.77_78insT
stem II
stem I
T-loop
stem III
3' stem loops
Nucleotide
UK Biobank all variants (rare and common)
b
Additional variants in individuals with NDD
Critical region
Allele count
n.63T>G
n.64_65insT, n.64_65insG, n.65A>G
n.67A>G
n.69C>T, n.68_69insA
n.76C>T
n.76_77insT
n.77_78insT, n.78A>C
T-loop
stem III
Confirmed de novo
Unknown/inherited
c
ACAGAGA sequence
U6
stem III
stem II
stem I
5'
3'
n.64_65insT
U4
3' stem loops
d
A77
C76
A78
C75
C74
n.64_65insT
A65
G64
U63
U62
U4
Quasi-
pseudoknot
A66
A67
U6
ACAGAGA
A68
U70
C69
U4/ U6
Stem I
U6
snRNA
U4
snRNA