Supplementary Information for:

Mutation spectrum of *NOD2* reveals recessive inheritance as a main driver of Early Onset Crohn's Disease

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Figure S1. Example pedigrees for homozygous and compound heterozygous *NOD2* carriers in an early onset IBD cohort. Example pedigrees *NOD2* variants identified in families with pediatric onset IBD. Diagnosis and age at diagnosis are indicated for the affected probands (filled dark grey symbols); unaffected relatives are depicted as unfilled symbols.

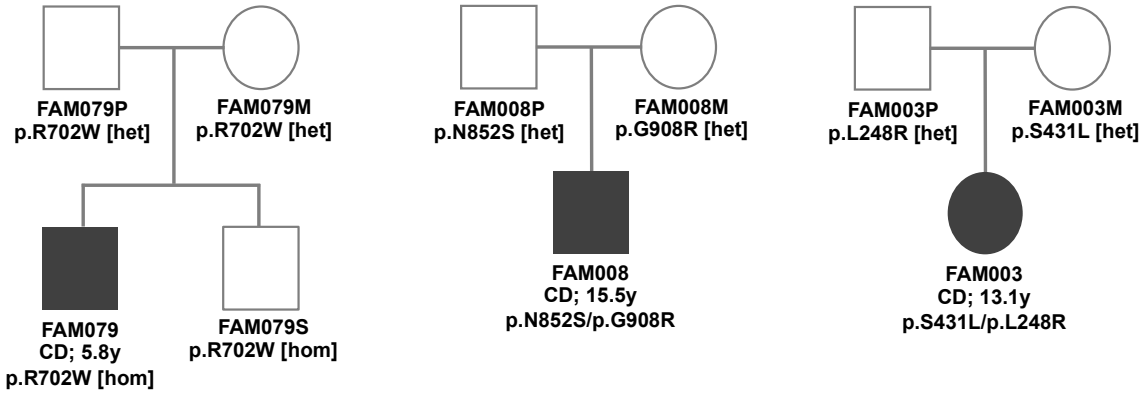

Figure S2. Example pedigrees for homozygous and compound heterozygous *NOD2* carriers in the RGC-GHS DiscovEHR cohort. Diagnosis indicated for the affected probands (filled dark grey symbols); unaffected relatives are depicted as unfilled symbols; individuals not sequenced are depicted as light grey symbols.

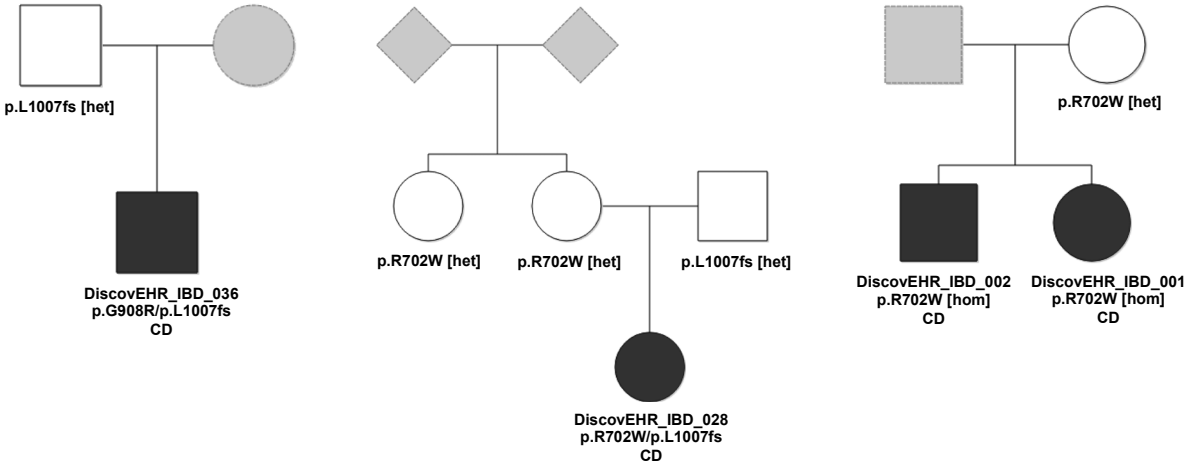

Figure S3. Exome sequencing coverage across the *NOD2* gene. Exon coverage and performance for all exons of the *NOD2* gene using the Nimblegen VCRome 2.1 target design.

Supplementary Tables:

Table S1. Clinical phenotypes of 92 early onset IBD probands carrying recessive *NOD2* variants.

Table S2. Clinical phenotype summaries for 92 early onset IBD probands carrying recessive *NOD2* variants.

Table S3. Clinical phenotypes of recessive *NOD2* carriers in the RGC-GHS DiscovEHR adult IBD cohort. Cases diagnosed ≤18y are highlighted in dark grey, and diagnosis ≤30y are highlighted in light grey.

Table S4. Genomic coordinates, annotation, frequencies, and bioinformatic prediction scores for *NOD2* variants identified in this study.