



Supplemental Fig. S3. Concordance plots LR-AS vs SR-GS

Concordance plot showing a comparison of EH5 genotypes from SR-GS compared to LR-AS sizing using Straglr for all RE loci, excluding those with solved cases which are presented in the main figures. Longer alleles are shown for loci associated with A. dominantly inherited. ataxias and B. other RE disorders. Both shorter and longer alleles are shown for C. X-linked recessive disorders. The locus specific pathogenic thresholds are shown as dashed red lines. A 1:1 correlation is shown as a dashed grey line. Individuals with no genotype call from either LR-AS, SR-GW or PCR are indicated separately to the numbered axis and are labelled nc (i.e. no genotype call). Triangles indicate low coverage (≤ 7 reads) on LR-AS and circles indicate samples with coverage > 7 reads for LR-AS.