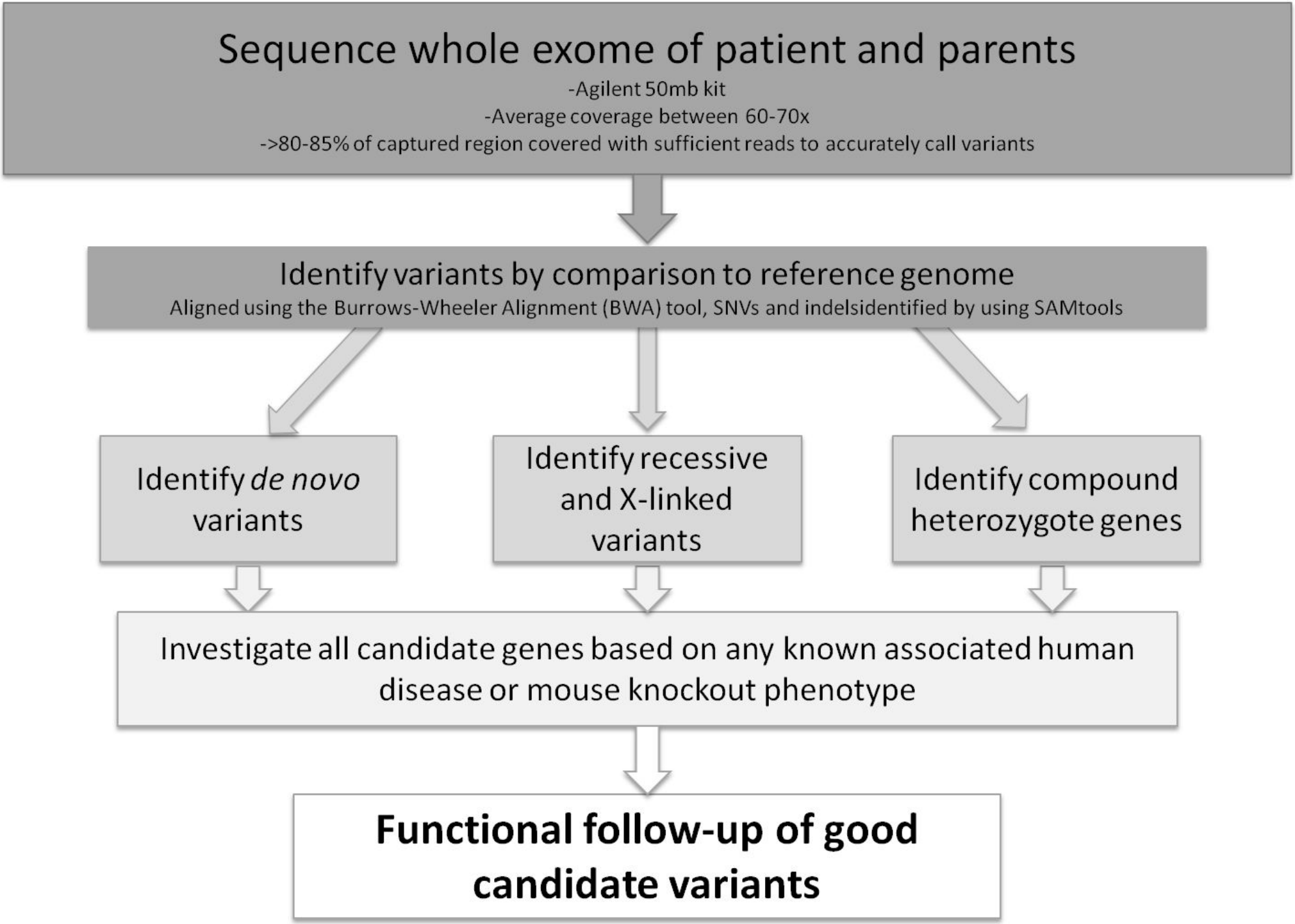
Sequence whole exome of patient and parents
-Agilent 50mb kit
-Average coverage between 60-70x
->80-85% of captured region covered with sufficient reads to accurately call variants
Identify variants by comparison to reference genome
Aligned using the Burrows-Wheeler Alignment (BWA) tool, SNVs and indelsidentified by using SAMtools
Identify *de novo* variants
Identify recessive and X-linked variants
Identify compound heterozygote genes
Investigate all candidate genes based on any known associated human disease or mouse knockout phenotype
Functional follow-up of good candidate variants