A clinical genetics centre treats Jenny, a patient with a family history of the RP10 form of autosomal dominant retinitis pigmentosa caused by a variant in the *IMPDH1* gene. The variant was initially found in her father some time ago and is described in a paper published just before he was diagnosed with the disease. The paper cites the GenBank RefSeq mRNA record NM_000883 when describing the structure of the gene. The variant is described using a nucleotide number from NM_000883 and a codon number from the translation product of that transcript. However, the version number of the GenBank record is not given in the paper and now, when the laboratory looks in GenBank (by following a hyperlink to NM_000883 given in the online version of the paper) they find that the current version is NM_000883.3, with a date stamp in March 2010. The exon structure of the gene was revised in 2003 and this resulted in the base, codon and exon numberings being changed. The variant reported in the literature is therefore no longer found at the expected location in the mRNA and protein sequences.

Laboratories specializing in this gene know that the numbering relative to the start codon has changed and recognize this as a potential source of error. Considerable effort is required to translate data in published papers and databases between different versions of reference sequences to gather the information needed to analyze cases like Jenny’s. This extra complexity means that the service may take longer and be more expensive than it otherwise might be. Unfortunately, new variants in the *IMPDH1* gene are still being described in the literature without specifying the version of the reference sequence.