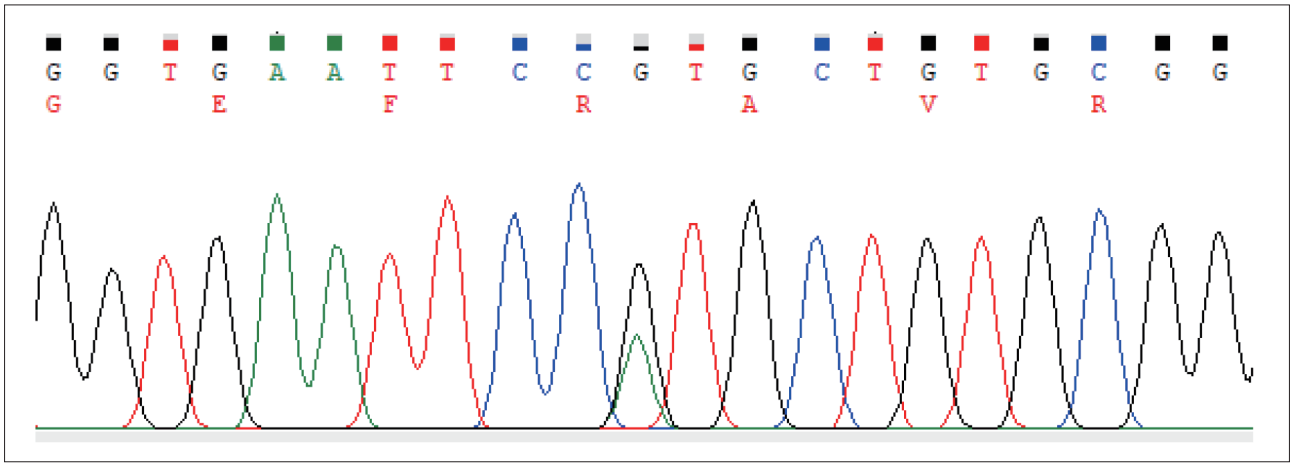

Supplementary Fig. 1. Sanger sequencing result of the proband showing a heterozygous missense mutation c.1871G>A (p.Arg624His, NM_004736.3) in the *XPR1*. This variant is considered likely pathogenic variants since it is in a well-established functional domain and a mutational hot spot, and it is not known as a benign variant (PM1, moderate evidence). Also, it is not included in control population databases such as Exome Sequencing Project, 1000 Genomes Project, or Exome Aggregation Consortium (PM2, moderate evidence). Lastly, multiple lines of in silico data support a deleterious effect on the gene or its product (PP3, supporting evidence). According to the American College of Medical Genetics and Genomics (ACMG) Standards and Guideline, this variant was concluded as likely pathogenic because it satisfied criteria (PM1, PM2, PP3) of the categories for classifying pathogenic variants.[1]

REFERENCES

1. Richards S, Aziz N, Bale S, Bick D, Das S, Gastier-Foster J, et al. Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. *Genet Med* 2015;17:405-424.