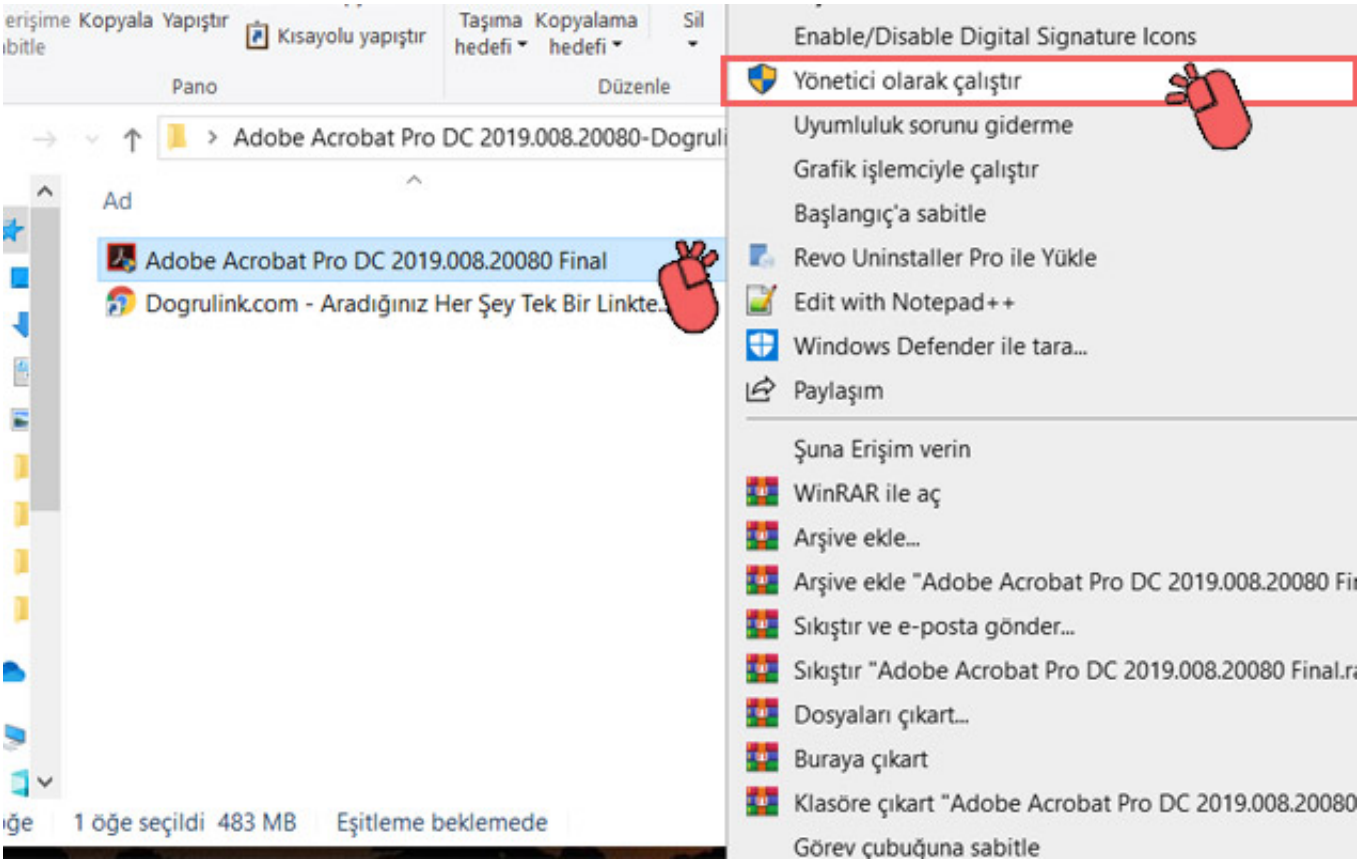
Soal Udkp Pns Polri



ssp1s2A point mutation in the human SLC26A4 gene causes anophthalmia and hearing loss. The genes involved in congenital aural, vestibular, and auditory ossicular malformation are not known. The objective of this study is to investigate the genetics of this complex disease. A two-year-old male infant with both congenital complete bilateral anophthalmia and severe sensorineural hearing loss (SNHL) was clinically diagnosed with Roberts syndrome. Clinical examination revealed anophthalmia, microtia, right ear canal atresia, and bilateral deafness. Using whole-exome sequencing, a point mutation (p.V329A) in the human solute carrier 26a4 (SLC26A4) gene was identified in this patient. SLC26A4 is highly expressed in the auditory organ, and mutations in this gene cause nonsyndromic hearing loss and variable clinical phenotypes. This point mutation is predicted to be pathogenic. The results of this study demonstrate that mutations in the SLC26A4 gene can cause anophthalmia. This observation further refines the genotype-phenotype correlation in anophthalmia. FILED NOT FOR PUBLICATION MAR 14 2012 MOLLY C. DWYER, CLERK UNITED STATES COURT OF APPEALS 520fdb1ae7