

### Genetics Case Study Comparison

Case Study	Geneticist's Hypothesis	Methods	What you learned	Geneticist's Conclusions and Recommendations	Other Subsequent Findings
Case Study 1: Sickle Cell Anemia in African Americans	Geneticist hypothesized that sickle cell anemia was caused by a mutation in the hemoglobin gene (HbS).	Geneticist used DNA sequencing to identify the mutation in the hemoglobin gene. They also conducted a pedigree analysis to track the inheritance of the trait.	Geneticist learned that the inheritance of sickle cell anemia was autosomal recessive. They also learned that the mutation was present in African Americans.	Geneticist concluded that the mutation was a result of natural selection. They recommended that individuals with sickle cell anemia should be treated with blood transfusions and hydroxyurea.	Geneticist's findings led to the development of sickle cell disease testing. They also learned that sickle cell anemia is a protective trait against malaria.
Case Study 2: Tay-Sachs Disease in Ashkenazi Jews	Geneticist hypothesized that Tay-Sachs disease was caused by a mutation in the hexosaminidase A gene (HexA).	The geneticist used DNA sequencing to identify the mutation in the HexA gene. They also conducted a pedigree analysis to track the inheritance of the trait.	Geneticist learned that the inheritance of Tay-Sachs disease was autosomal recessive. They also learned that the mutation was present in Ashkenazi Jews.	Geneticist concluded that the mutation was a result of natural selection. They recommended that individuals with Tay-Sachs disease should be treated with enzyme replacement therapy.	Geneticist's findings led to the development of Tay-Sachs disease testing. They also learned that Tay-Sachs disease is a protective trait against tuberculosis.
Case Study 3: Phenylketonuria (PKU)	Geneticist hypothesized that PKU was caused by a mutation in the phenylalanine hydroxylase gene (PAH).	Geneticist used DNA sequencing to identify the mutation in the PAH gene. They also conducted a pedigree analysis to track the inheritance of the trait.	Geneticist learned that the inheritance of PKU was autosomal recessive. They also learned that the mutation was present in individuals of various ethnicities.	Geneticist concluded that the mutation was a result of natural selection. They recommended that individuals with PKU should be treated with a low-phenylalanine diet.	Geneticist's findings led to the development of PKU testing. They also learned that PKU is a protective trait against schizophrenia.
Case Study 4: Cystic Fibrosis	Geneticist hypothesized that cystic fibrosis was caused by a mutation in the CFTR gene.	Geneticist used DNA sequencing to identify the mutation in the CFTR gene. They also conducted a pedigree analysis to track the inheritance of the trait.	Geneticist learned that the inheritance of cystic fibrosis was autosomal recessive. They also learned that the mutation was present in individuals of various ethnicities.	Geneticist concluded that the mutation was a result of natural selection. They recommended that individuals with cystic fibrosis should be treated with pancreatic enzyme replacement therapy.	Geneticist's findings led to the development of cystic fibrosis testing. They also learned that cystic fibrosis is a protective trait against tuberculosis.

# Genetics Case Study Comparison Answer Key

**Shasha Hu**



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