

Revised Prevalence Estimate of Possible Hereditary Xerocytosis as Derived from a Large US Laboratory Database

Background

- Hereditary xerocytosis (HX) is a rare form of hemolytic anemia. The disease is caused by mutations in either of the 2 genes that encode ion-channel proteins in the membrane of red blood cells.¹
- HX prevalence in the United States was estimated to be 1 in 50,000 births, but these estimates are based on small numbers of reported cases.¹ Recent studies suggest prevalence may be higher.^{2,3}
- **Objective:** To better define the prevalence of HX, the investigators examined test results of complete blood counts (CBCs) available from a large database at a US commercial laboratory database.

Methods

- Results of tests performed between January 2014 and December 2016 were retrieved from the Quest Diagnostics Health Trends™ Database.
- These results included all CBC tests that contained mean corpuscular hemoglobin concentration (MCHC), hemoglobin, and mean corpuscular volume (MCV).
- People were categorized as follows:
 - Possible HX with anemia: MCHC levels above the reference range (RR) and hemoglobin levels below the RR
 - Possible HX with compensated hemolysis: MCHC levels above the RR, hemoglobin levels within or above the RR, and elevated total bilirubin, ferritin, lactate dehydrogenase, or reticulocyte counts
- The prevalence of possible HX was estimated by extrapolation of the results to the 2010 US Census adult population.
- A chi-square test was used to determine statistical significance between groups.

Results

- The study group included more than 48 million people; 59.1% were female.
- People with test results that fell into the study categories included:
 - Possible HX with anemia: 2,272 (0.008%) females and 1,757 (0.009%) males
 - Possible HX with compensated hemolysis: 358 (0.002%) females and 1,651 (0.010%) males
- The prevalence of possible HX with anemia and possible HX with compensated hemolysis in the United States was estimated to be about 1 in 7,000 adults.

Conclusions

- The estimated prevalence of possible HX from this study is about 7 times greater than that previously reported.
- These patients may be candidates for screening for mutations associated with HX: those with possible HX with anemia and those with possible HX with compensated hemolysis.

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Webpage

<https://www.ncbi.nlm.nih.gov/pubmed/?term=28971500>

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