What Is the Impact of Direct-to-Consumer (DTC) Genetic Testing in the Diagnosis of Alpha-1 Antitrypsin Deficiency (AATD)?

**STUDY DESIGN**

- Cross-sectional survey of DTC recipients of reactions and reported health behavior responses
- 195,014 individuals recruited from all genotyped 23andMe customers
- The two loci used to determine genetic risk for AATD were rs28929474 (PI*Z) and rs17580 (PI*S)

**RESULTS**

- 27% of those with physician-diagnosed AATD reported first becoming aware of AATD through the DTC test
- DTC testing results were associated with reported decreases in alcohol consumption (OR 4.0; 2.6-5.9) and cigarette smoking (OR 1.7; 1.4-2.2)

### Allele Variant Frequency

\[
\begin{align*}
6.5\% & \text{ PI*Z} \\
15.1\% & \text{ PI*S} \\
0.63\% & \text{ PI*ZZ} \\
21.6\% & \text{ PI*ZZ}
\end{align*}
\]

DTC testing aided in the identification of at-risk individuals and families and the information in the personalized AATD report prompted positive self-reported behavior change.


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