Understanding your results

This test is used to determine alpha tryptase copy number. Alpha tryptase is encoded by the TPSAB1 gene which is on chromosome 16. The primary gene products of this gene are β-tryptase, which can be produced by TPSAB1 and TPSB2, and α-tryptase, which is only produced by the TPSAB1 gene. Individuals who inherited one copy of α-tryptase–encoding sequence on both have normal serum tryptase levels.

This test measures both α- and β-tryptase. A normal result is any combination of α- and β-tryptase that adds up to 4. A positive result – which means there is a duplication or triplcation of α-tryptase – is a combination of α- and β-tryptase that is 5 or higher. An explanation of the results are included below, however there may be other copy number changes in these genes that have not yet been described. In addition, the assay cannot distinguish between the possible 4,2 genotypes without testing of additional family members.

NORMAL RESULT: Alpha-tryptase copy number 2; beta-tryptase copy number 2

![Diagram](TPSB2) ![Diagram](TPSAB1)

NORMAL RESULT: Alpha-tryptase copy number 1; beta-tryptase copy number 3

![Diagram](TPSB2) ![Diagram](TPSAB1)
NORMAL RESULT: Alpha-tryptase copy number 0; beta-tryptase copy number 4

TPSB2 --- TPSAB1
β --- β
β --- β

POSITIVE RESULT: Alpha-tryptase copy number 2; beta-tryptase copy number 3

TPSB2 --- TPSAB1
β --- α --- α
β --- β

POSITIVE RESULT: Alpha-tryptase copy number 3; beta-tryptase copy number 2

TPSB2 --- TPSAB1
β --- α --- α
β --- α
POSITIVE RESULT: Alpha-tryptase copy number 4; beta-tryptase copy number 2. There are two possibilities for a 4,2 result, although the first type is the most common.