

CCR5 Delta 32 GENETIC TESTING

Department of Molecular Biology

Background

For HIV to infect human cells, certain proteins called 'co-receptors', must be present on the cellular surface. These proteins are made in human cells according to the genetic information contained within the individual's chromosomes. One particular protein, called the CCR5 co-receptor, is present on the surfaces of CD-4 lymphocytes and is particularly important in the early stages of HIV-1, the most common form of HIV infection within India.¹

Every human has two CCR5 genes; one is inherited from each parent.

Some studies have suggested that for some people who have certain mutations in the gene that encodes for the CCR5 protein have a significantly reduced risk of becoming infected with HIV.²

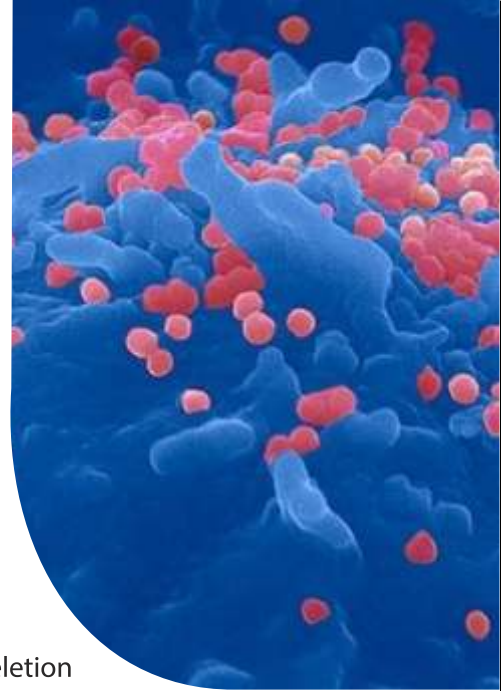
"CCR5-delta32" is a deletion mutation of a gene which only 1% of the total population has two copies of this gene and individuals who carry two copies of this genetic mutation are immune to Smallpox, The Bubonic Plague (Black Death) and resistant to HIV, the virus that causes AIDS. Up to 20% of the population carry only one copy of this genetic mutation depending on your background and although they still run a significant risk of contracting HIV, the progress of the disease is greatly reduced and can result in a longer life expectancy.

What is CCR5-delta32?

Cysteine-cysteine chemokine receptor 5 (CCR5) is found in the cell membranes of many types of mammalian cells, including nerve cells and white blood cells. The role of CCR5 is to allow entry of chemokines into the cell—chemokines are involved in signaling the body's inflammation response to injuries. The gene that codes for CCR5 is situated on human chromosome 3. Various mutations of the CCR5 gene are known that result in damage to the expressed receptor. One of the mutant forms of the gene is CCR5-delta32, which results from deletion of a particular sequence of 32 base-pairs. This missing information results in a co-receptor with a slightly different shape on the surface of the CD-4 lymphocyte. This mutant form of the gene results in a receptor so damaged that it no longer functions. But surprisingly, this does not appear to be harmful.

Why CCR5-delta32 is Resistant to HIV?

This mutation can be advantageous to those individuals who carry it. The virus HIV normally enters a cell via its CCR5 receptors, especially in the initial stage of a person becoming infected. But in people with receptors crippled by the CCR5-delta32 mutation, entry of HIV by this means is blocked, providing resistance to HIV for people with delta 32 mutation on both genes (called homozygous carriers) and greatly slowing progress of the disease in people with a delta 32 mutation on one of the two genes (called heterozygous carriers).



Methodology: Polymerase Chain Reaction (PCR)

Turnaround Time: 1-2 working days

Specimen Type: EDTA – Blood (Purple top tube) – 4 ML

Result:

A result of “**Wild type**” means that neither copy of your CCR5 gene has the deletion; you do not carry the deletion.

A result of “**Heterozygous**” means that one copy of your CCR5 gene has the deletion and the other does not. For those viruses affected by CCR5 there is an increased resistance but not immunity.

A result of “**Homozygous**” means that both copies of your CCR5 gene have the deletion and the cell receptor controlled by CCR5 is not present. This may provide immunity from certain viruses which use that receptor to identify and attack cells.

References:

1. Oelrichs R. (2004) The Subtypes of human immunodeficiency virus in Australia and AISA. CSIRO Journal of Sexual Health;1:1-11
2. Marmor M, Sheppard HW, Donnell D, et al. (2001) Homozygous and Heterozygous ccr5-DELTA]32 Genotypes Are Associated With Resistance to HIV Infection. Journal of Acquired Immune Deficiency Syndromes; 27:472-481.



Unipath
SPECIALTY LABORATORY Ltd.

Unipath Specialty Laboratories Ltd.

102, Sanoma Plaza, B/S JMC House, Opp. Parimal Garden, Ellis Bridge, Ahmedabad - 380006

Phone No. 079-49006800/31

Mail to: unipathmdx@gmail.com