Correlation Between *DEFB104* Gene Polymorphisms and HIV-1 Vertical Transmission in Brazilian Children

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Although significant developments have been achieved to improve prevention and effective treatment against AIDS, the incidence of HIV-1 infection and deaths due to AIDS keeps growing in underdeveloped countries. The HIV-1 vertical transmission (VT) from mother to child is a complex interplay involving environmental, viral and host factors. Defensins, a family of antimicrobial peptides, play an important role against viral infection on innate immunity and previous findings described that beta-defensins disrupt HIV-1 membrane. Beta-defensins have great potential as molecular targets to study the susceptibility to HIV-1 infection, especially in newborns, in which acquired immunity response is not fully developed. The aim of this study was to investigate the association between functional copy number polymorphisms (CNPs) of human beta-defensin 4 gene (*DEFB104*) and susceptibility to HIV-1 VT in Brazilian children. We enrolled 36 children born from HIV-positive mothers that contracted infection during delivery (HIV-positive), 24 non-infected children born from HIV-positive mothers (exposed-uninfected) and 36 children of HIV negative mothers (healthy controls). *DEFB104* CNP assessment was done with the Multiplex Ligation-dependent Probe Amplification (MLPA) technique (*MRC-Holland*). The average of *DEFB104* gene CNPs was significantly lower in HIV-positive children when compared with exposed-uninfected children *($p=0.006$).* Lower copy number of *DEFB104* gene produces low levels of hBD-4 proteins and can explain in part ours findings. Thus, despite the restricted number of samples, our results suggest that *DEFB104* gene is involved in protecting from HIV-1 VT but require further investigation.

Key words: *DEFB104, HIV-1, CNPs, MLPA*

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