

Innate Immune Dysfunction in Niemann Pick Disease Type C

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Abstract : Niemann-Pick Type C disease is a rare, usually fatal lysosomal storage disorder. Although clinically characterized by progressive neurodegeneration, there is also evidence of altered innate immune responses such as neuroinflammation that promote disease progression. We have initiated an investigation into whether phagocytosis, an important innate immune activity and the process by which particles are ingested is defective in NPC. Using an in vitro assay, we have shown that NPC macrophages have a deficiency in the phagocytosis of different particles. We plan to investigate the mechanistic basis for impaired phagocytosis, the contribution that this deficiency makes to disease pathology, and whether therapies that have shown in vivo benefit are able to restore phagocytic activity.

Keywords : Niemann Pick Disease C, phagocytosis, innate immunity, lysosomal storage disorder

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