

The Feasibility Of Using Cryopreserved Lymphoblastoid Cells To Diagnose Some Lysosomal Storage Diseases

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The Epstein-Barr virus (EBV) is utilized as a tool in the study of cellular biology because of its capacity to transform the B-Lymphocytes. For this reason, EBV is used in the conservation of human B-Lymphocytes for long periods for subsequent evaluation of the lysosomal hydrolase activity. We present biochemical data in this paper that demonstrates the validity of lymphoblastoid cell lines for the diagnosis of GM1-gangliosidosis, Gaucher, Fabry and Pompe diseases, and Mucopolysaccharidosis type I. This study was performed in cultures of peripheral blood and the investigations included analysis of normal subjects (25 cases) by measurement of the enzyme activities and immunohistochemistry (IHC). The activities of the enzymes β -galactosidase, β -glucosidase, α -iduronidase, α -galactosidase and α -glucosidase were measured before and after cryopreservation (180 days). When the transformation was confirmed by IHC we measured the enzymatic activity. We observed some significant alterations in the enzymatic activity of the non-cultivated cells when we compared them to others that had been in culture for 12 days and held frozen for 180 days. However, these alterations do not invalidate the use of the technology of transformation of the lymphoblastoid cell lines with EBV to diagnose the diseases mentioned above in view of the fact that the cultivated cells, before and after freezing, demonstrated similar enzymatic activities.

Key-words: Epstein-Barr virus, lymphoblastoid cell lines, lysosomal hydrolases, cryopreservation.

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