Dihydrolipoamide Dehydrogenase Deficiency in Southern Israel:

A rare disease with a change of natural history

Pyruvate dehydrogenase complex (PDHC) is composed of 3 components: E1 which is the rate limiting enzyme, E2 and E3. E3 is a flavoprotein - Dihydrolipoamide Dehydrogenase ( DLD E3; MIM \* 238331; EC 1.8.1.4) is common to all 2-ketoacid dehydrogenases. Defects in the PDHC leads to abnormal conversion of pyruvate into lactate instead of acetyl –CoA through the TCA cycle.

Deficiency of PDHC interferes with energy production from glycolysis. Therefore PDHC deficiency, and DLD- E3 deficiency as part of it, aggravates lactic acidosis by carbohydrates consumption.

DLD- E3 is a rare autosomal recessive inherited disorder. Approximately 28 cases have been reported worldwide. Consanguinity increases the frequency. Over the last 20 years more than 10 cases have been reported in Southern Israel.

The gene for DLD- E3 is located on 7q31-q32. So