**Univerzita Karlova v Praze, Přírodovědecká fakulta**

Katedra organické a jaderné chemie
zve všechny zájemce na přednášku z cyklu

# Quo Vadis Chemie

## Tandem Mass Spectrometry in Clinical Enzymology: Towards Newborn Screening for Mucopolysaccharidoses and Other Lysosomal Storage Diseases

kterou přednese

## Prof. František Tureček

Department of Chemistry, University of Washington, Seattle, WA, USA

**dne 23.3. 2010 v 10:00 hod.**
**v posluchárně CH2, v budově chemických kateder PřF UK**
**Hlavova 8, Praha 2**

Abstrakt:

*Screening for rare metabolic diseases of entire newborn populations has been extending in the U.S. over the last decade or so. Screening is done for diseases for which there are preventive or medicinal therapies, as pioneered for phenylketonuria in the 1960's. Including new diseases into newborn screening protocols requires new technologies to be developed such as tandem mass spectrometry. Our approach relies on specific and quantitative mass spectrometric assays of several enzymes in catabolic degradation pathways in the lysosome. Examples will be given for enzyme assays pertinent to the group of lysosomal storage diseases, mucopolysaccharidoses I, II, III, IV, and VI, that degrade complex polysaccharides such heparan, dermatan, keratan, and chondroitin sulfate. Saccharide enzyme substrates are designed and synthesized by procedures that allow them to be scaled to multigram quantities for global supply. In addition to the sugar moiety which is recognized by the enzyme, the substrates also contain linkers or groups for the introduction of stable isotopes into internal standards, and functionalities favorably affecting solubility, solid-liquid or liquid-liquid partitioning, and mass spectrometric fragmentations.*