

NATIONAL METABOLIC BIOCHEMISTRY NETWORK

HST STUDY DAY: JULY 17th 2006

Lysosomal Storage Diseases/Neuronal Ceroid Lipofuscinoses

The MetBioNet HSTs gathered at the Institute of Child Health (Great Ormond Street Hospital) on July 17th for a first specialist study day designed as a specific part of their training programme.

The morning session was devoted to clinical and laboratory aspects of the lysosomal storage diseases and commenced with a masterly presentation from Professor Bryan Winchester (Endocrine & Metabolism Unit – ICH) who reviewed our current understanding of this group of disorders and their clinical manifestations. He reminded the group that the list of diseases continues to grow and extends beyond deficiencies of luminal lysosomal enzymes to include problems arising from lack of protein co-factors and membrane transporters, plus defects of post-translational processing in all these groups. He brought the story up to the present day by indicating the current status of enzyme replacement therapy and its monitoring plus other postulated treatment options for selected disorders.

Marie Jackson (Enzyme Laboratory – GOSH) then reviewed the analytical approaches required (and their pitfalls) for the necessary range of lysosomal enzymes, emphasising that a range of substrate labels still needs to be employed. An awareness of the associated pre-analytical problems in providing the service is critical and detailed clinical information from referring laboratories is helpful in prioritising the enzyme panels tested. Collaboration with other diagnostic departments was seen as an essential part of arriving at a diagnosis in complex and atypical cases.

The importance of a collaborative approach was reinforced during the afternoon session, which focussed on the laboratory diagnosis of the Neuronal Ceroid Lipofuscinoses. Speakers from histology/electron microscopy (Glenn Anderson), enzymology (Marie Jackson) and molecular genetics (David Cregeen) each outlined their contribution to diagnosing this group of disorders. Together they highlighted how knowledge from several disciplines continues to aid our understanding of these conditions and attempts at their scientific classification.

There was plenty of time during the day for questions and discussion within the small group and informal feedback from attendees has been positive in respect of both the format and content of the day.

A large vote of thanks must go to Marie Jackson for both her scientific and organisational contribution to the meeting that certainly got this dimension to the MetBioNet HST training programme off to an exciting start.

Colin Samuell (27.7.06)