

ERNDIM common sample 2007

JR Bonham, Sheffield



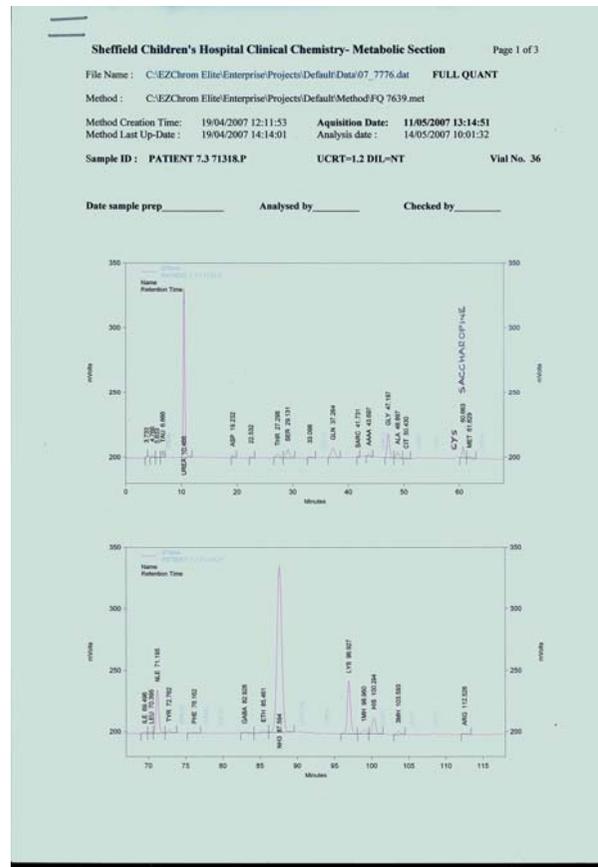
The Sample

- **Patient**

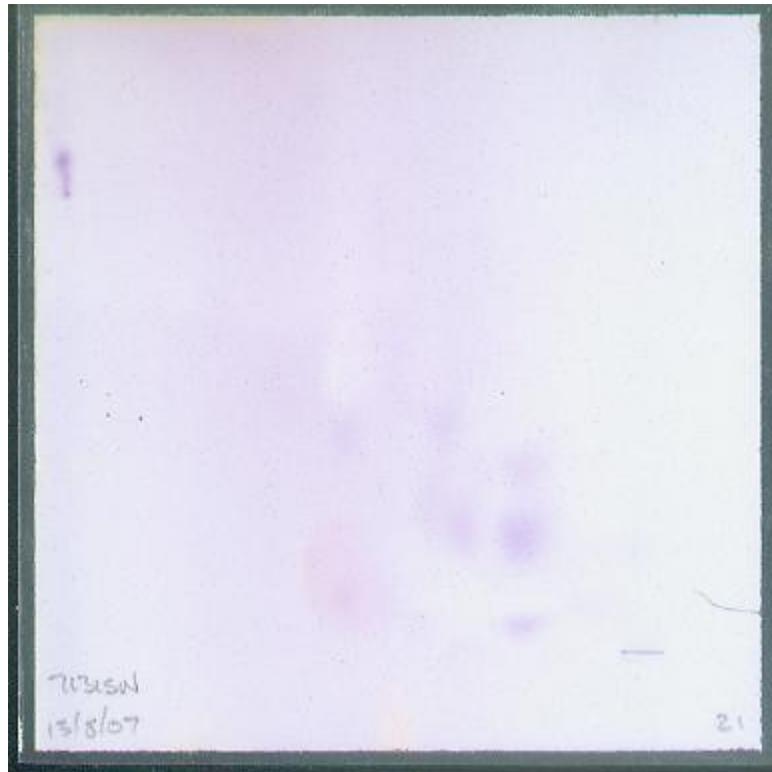
A female, aged 18 years, learning difficulties and behavioural problems since 5 years of age. She also has short stature

This sample was obtained from an 18 year old woman with saccharopinuria

The findings



The findings



The results of the survey

	Sheffield	Prague	Lyon	Basel	Maastricht
Number responding	20/21	17/18	22/22	20/21	19/19
Increased lysine	15/20	17/17	19/22	19/20	17/19
Mean lysine ($\mu\text{mol/L}$)	291	271	257	252	240

The results of the survey

	Sheffield	Prague	Lyon	Basel	Maastricht
Saccharopine identified	6/20	7/17	13/22	7/19	6/19
α-aa sas defn hyperlysinaemia I or II hyperlysinuria	8/20	14/17	16/22	11/19	12/19
Lysinuric protein intolerance	4/20	0/20	4/22	3/19	1/19

The results of the survey

	Sheffield	Prague	Lyon	Basel	Maastricht
No significant abnormality	4/20	0/17	3/22	5/19	4/19
Some other disorder	0/20	2/17	5/22	1/19	1/19
Plasma aminoacid analysis advised	15/20	13/17	15/22	12/19	11/19
Enzymic/molecular genetic confirmation advised	1/20	12/17	14/22	12/19	9/19

Scoring

- Analytical performance
 - Noting an increased excretion of lysine
Scored 1 point
 - Noting excretion of saccharopine
Scored 1 point

Scoring

- Interpretative proficiency
 - α -amino adipic semialdehyde deficiency, Hyperlysinaemia, hyperlysinaemia type 1, hyperlysinaemia type 2, saccharopinuria, hyperlysinuria
Scored 2 points
 - Lysinuric protein intolerance
Scored 1 point
 - No diagnostic abnormality, anything else
Scored 0 points

Scoring

- Recommendations
 - Plasma aminoacid analysis
 - Enzyme/mutational confirmation

Scored 1 point alone or in combination

Average Scores

	Sheffield	Prague	Lyon	Basel	Maastricht
Average score of responders	2.95	4.2	3.86	3.63	3.10