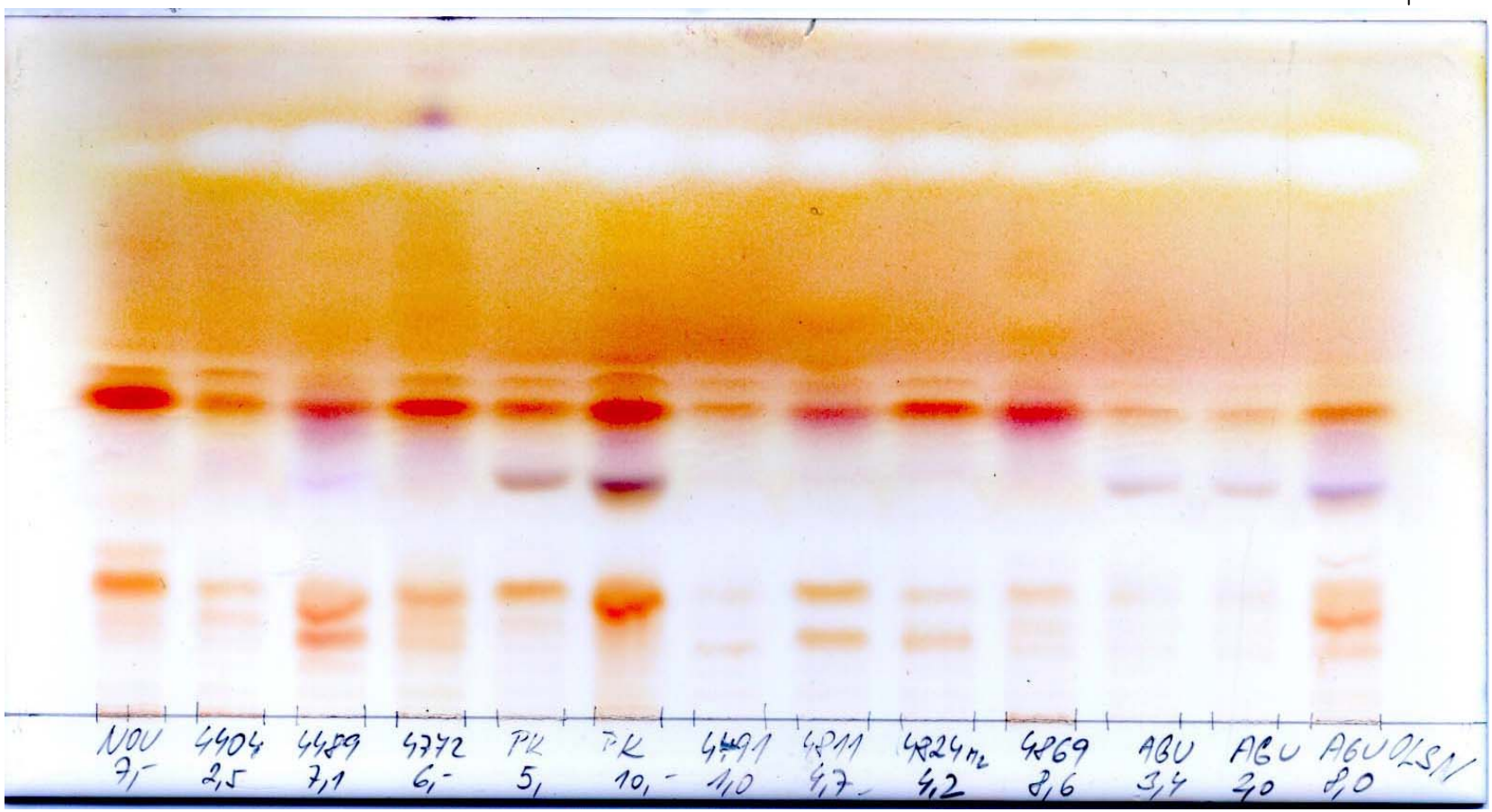
LSD in DPT Schemes



Scoring system

A	**Analytical performance**	Correct results of the appropriate tests	2
		Partially correct or non-standard methods	1
		Unsatisfactory or misleading	0
I	**Interpretative proficiency**	Good (diagnosis was established)	2
		Helpful but incomplete	1
		Misleading/wrong diagnosis	0
R	**Recommen-dations**	Helpful	1
		Unsatisfactory or misleading	0

Samples in DPT Centers

- DPTC Amsterdam n/a
- DPTC Basel
  - 2006 GM1 gangliosidosis
- DPTC Lyon
  - 2003 sialidosis
  - 2004 fucosidosis
  - 2005 alpha-mannosidosis
- DPTC Prague
  - 2001 alpha-mannosidosis
  - 2002 MPS VII
  - 2003 Sialidosis
  - 2004 MPS III
  - 2005 MPS II
  - 2006 Aspartylglucosaminuria
- DPTC Sheffield n/a

Samples in DPT Centers

Diagnosis	A	I	R	T	MISSED
A-Mann PRG	79	74	69	74	5/21
MPS VII	65	75	70	69	8/20
Sialidosis-P	72	75	89	77	3/20
MPS III	38	28	55	37	11/20
GM1	65	55	80	66	8/20
A-Mann Lyon	73	73	64	71	1/22
Fucosidosis	66	66	84	69	6/20
Sialidosis-L	50	32	58	44	11/20
MPS II	73	75	79	75	1/24
AGU	64	61	72	64	8/19

A- aspartylglucosaminuria

- Analytical performance 64%
  - AA 5/18 detected AGU
  - OLS 9/11 AGU
- Interpretative proficiency 61%
  - 7/18 missed diagnosis
- Recommendations 72%

A- aspartylglucosaminuria

Amino acids n=18	
Aspartylglucosamine	3
Unidentifiable peak coeluting with urea – aspartylglucosamine?	1
1-D-ELFO shows a brown fraction near to origin (acidic). 2-D-electro/chromat shows grey / blue fraction. After acid hydrolysis aspartic acid increased. Suggestive of aspartylglucosamine	1

Oligosaccharides n=11	
**Pattern typical for AGU	9
*OLS, pathological pattern resembling Morbus Schindler	1
OLS non specific – mildly elevated	1

A- aspartylglucosaminuria

**Aspartylglucosaminuria	11
No diagnosis	4
Morbus Schindler. Differential diagnosis: sialidosis	1
Salla disease. Hypophosphatasia.	1
Possible adenylsuccinase deficiency	1
No answer	1

***Enzyme assay / Aspartylglucosaminidase in leucocytes/fibroblasts**	**11**
***Mutation analysis**	**10**
***OLS analysis / control urine for OLS**	**2**