Newborn screening in Sweden

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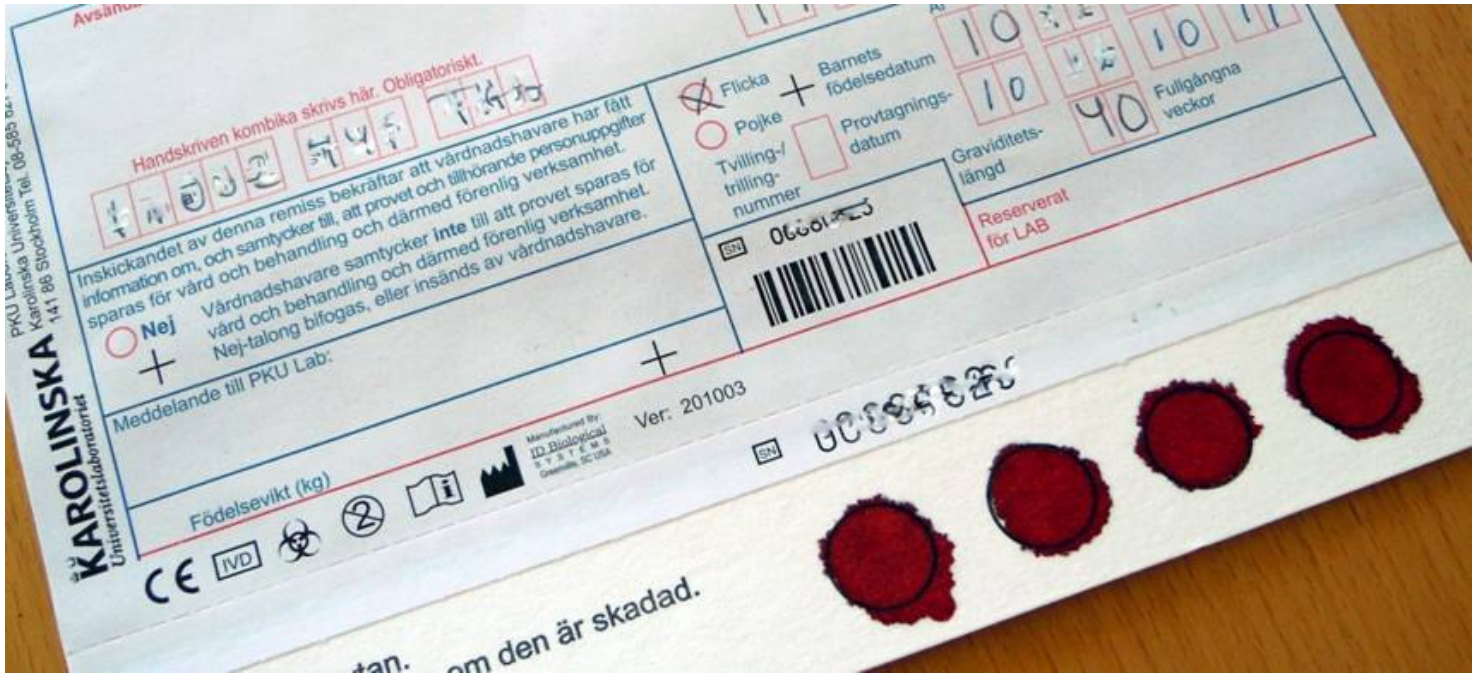


History

Start 1965 with PKU, DBS

Five disorders by 2010

From Nov 15, 2010,
expansion with 19 disorders

The screening laboratory

One centralized lab, presently 110 000 births/year

State bacteriological laboratory 1965 (Palmstierna, Larsson, Hagenfeldt)

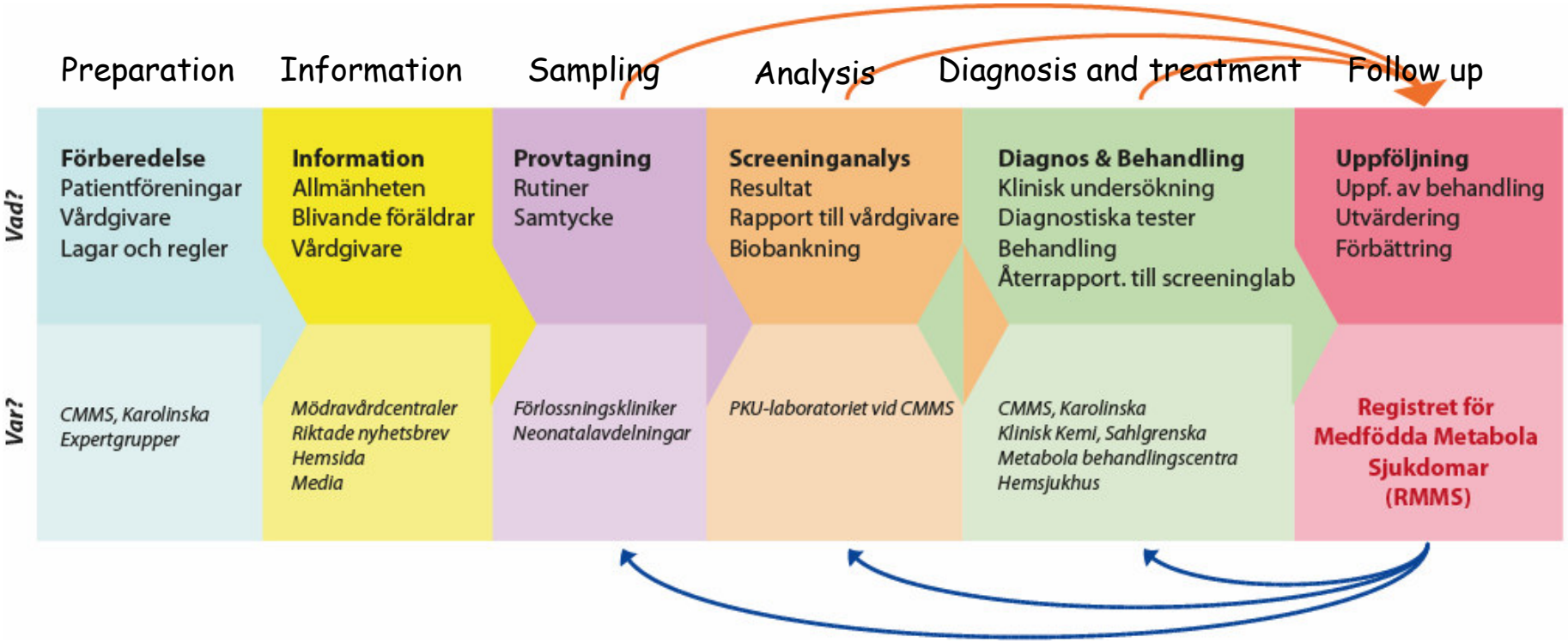
Merged with the diagnostic lab for inborn errors of metabolism in Huddinge Stockholm, 1998, to create *Centre for Inherited Metabolic Diseases, CMMS*

Moved to Karolinska Solna, 2011

Accumulated results, 1965 - 2011

	Number screened	True pos	PPV% 1965-2011	PPV% 2007-2011
PKU 1:16 300	4 600 000	283	36	61
GALT 1:105 000	4 500 000	43	9	67
CH 1:2 800	3 300 000	1170	73	74
CAH 1:11 800	2 720 000	230	13	10
BIOT 1:69 600	974 000	14	45	36

Newborn screening
- from preparation to follow up



Lene Sörensen, CMMS

Process for evaluation disorders to be included

There has not been a defined process

The National Board of Health and Welfare has initiated work to establish a process

Criteria for the evaluation have been defined

Applications will be subject to an HTA, followed by evaluation by an expert group and finally a reference group will give a recommendation to the head of the National Board of Health and Welfare, who will decide

Information

Homepage

Written information, 3rd trimester, posters

Courses for midwives 2-4 times/year

Oral and written information at sampling

Sampling

As soon as possible after 48 hrs of age.

To a community post box same day.
The screening lab sends out stamp equivalents to support this.

The average age at sampling is now 2,9 days and arrival in the lab at age 5,5 days.

Screening lab

Lab results ready next day at the latest

A physician in the lab phones treatment centre (or local pediatric clinic)

Written report is sent

Normal results are sent to delivery clinics regularly

Five specialized centres for diagnosis and treatment of IEM

Umeå, Uppsala, Stockholm, Göteborg, Lund

Teams with dietitians, nurses, psychologists, specialized paediatricians, home health care and habilitation teams

Metabolic centre

Clinical investigation

Diagnostic samples

Second DBS

Treatment

Report to screening lab – including long term outcome

RMMS, registry for inborn errors of metabolism

A registry for the follow up of treatment is built at the screening lab in cooperation with the treatment centres

Built on a platform that is used by other Swedish registries – the "childrens platform"

All 20 disorders screened by MS-MS are to be included to start with

Screening programme

Fatty acid degradation defects:

- CUD
- CPT1
- CPT2
- CACT
- MCAD
- VLCAD
- LCHAD, TP
- GA2 (MAD)

Organic acidurias:

- MMA (cbl, B12-brist)
- PA
- IVA
- BKT
- GA1

Urea cycle disorders:

- Citr1,2
- ASA
- ARG

Miscellaneous:

- Tyr1
- MSUD
- Homocystinuri

The earlier:

- PKU
- CH
- CAH
- GALT
- BIOT

The Region 4 collaborative

Before we started the expanded screening we could join the R4 project for screening led by Professor Piero Rinaldo at the Mayo Clinic

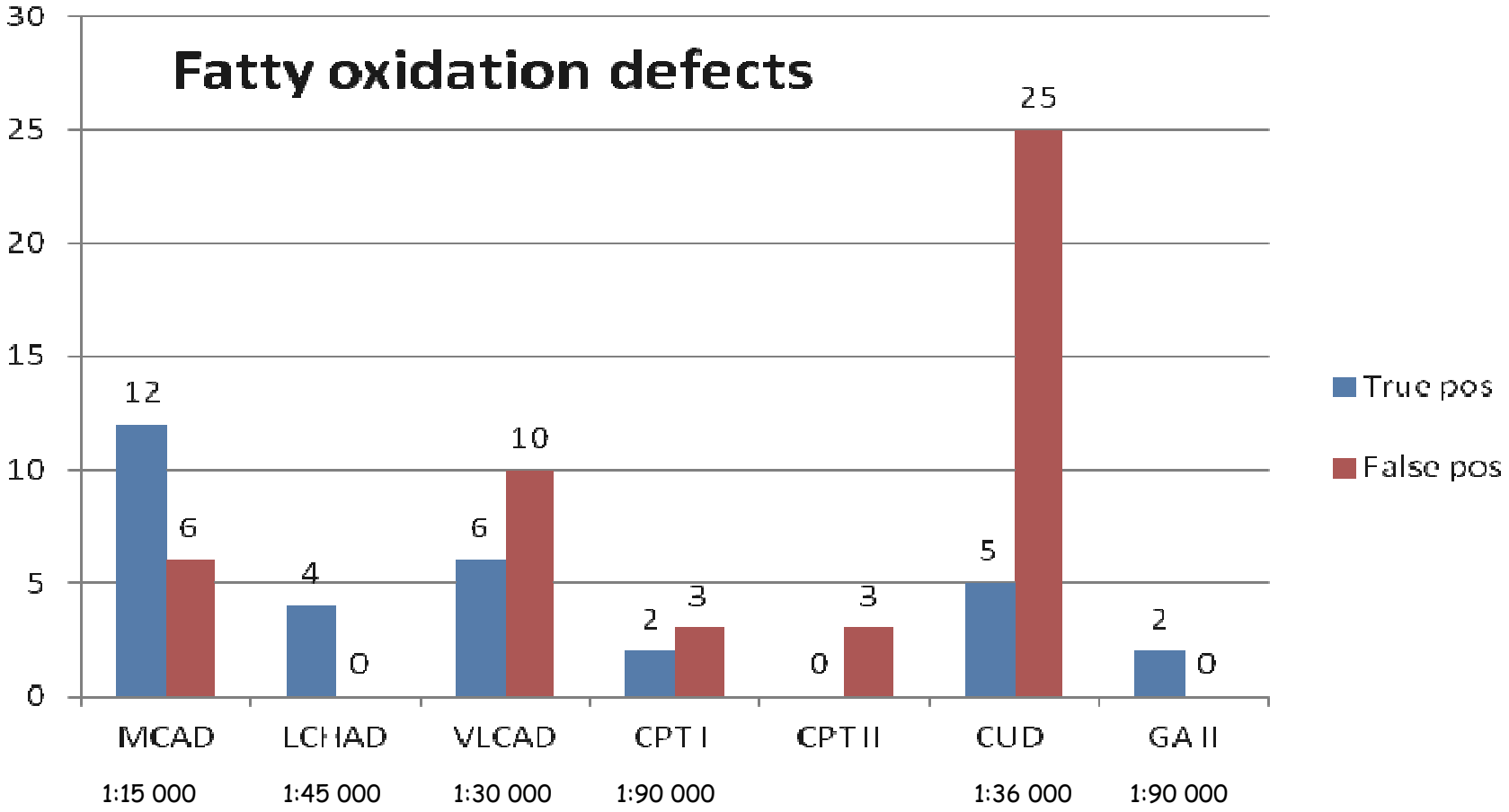
Two of us attended a one week course led by him prior to the start

Another four of us have attended the course and we have used the on-line tools developed at Mayo from the very start

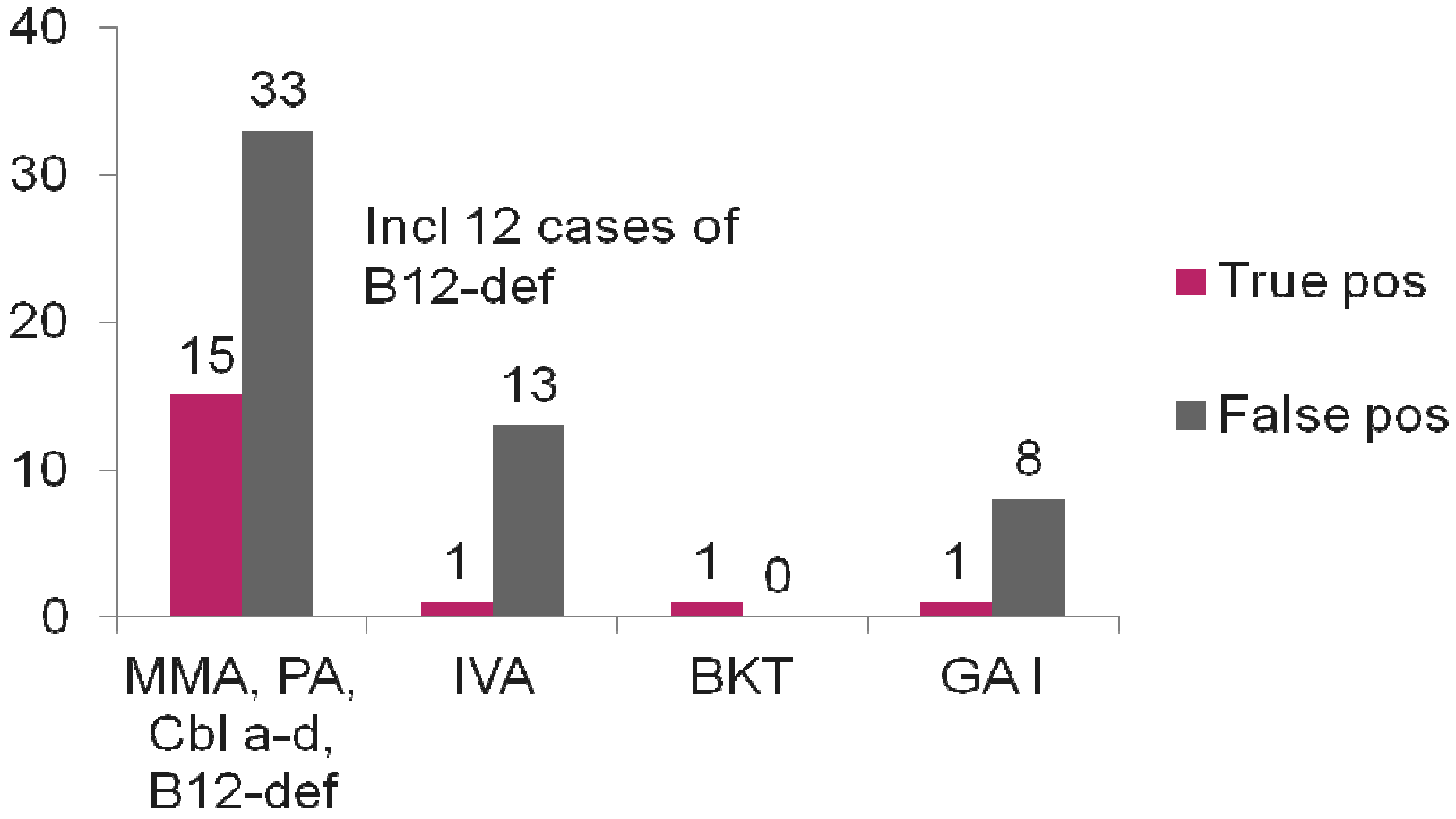
This has been of paramount importance for the success of our program

Results nov 15, 2010-june 30, 2012

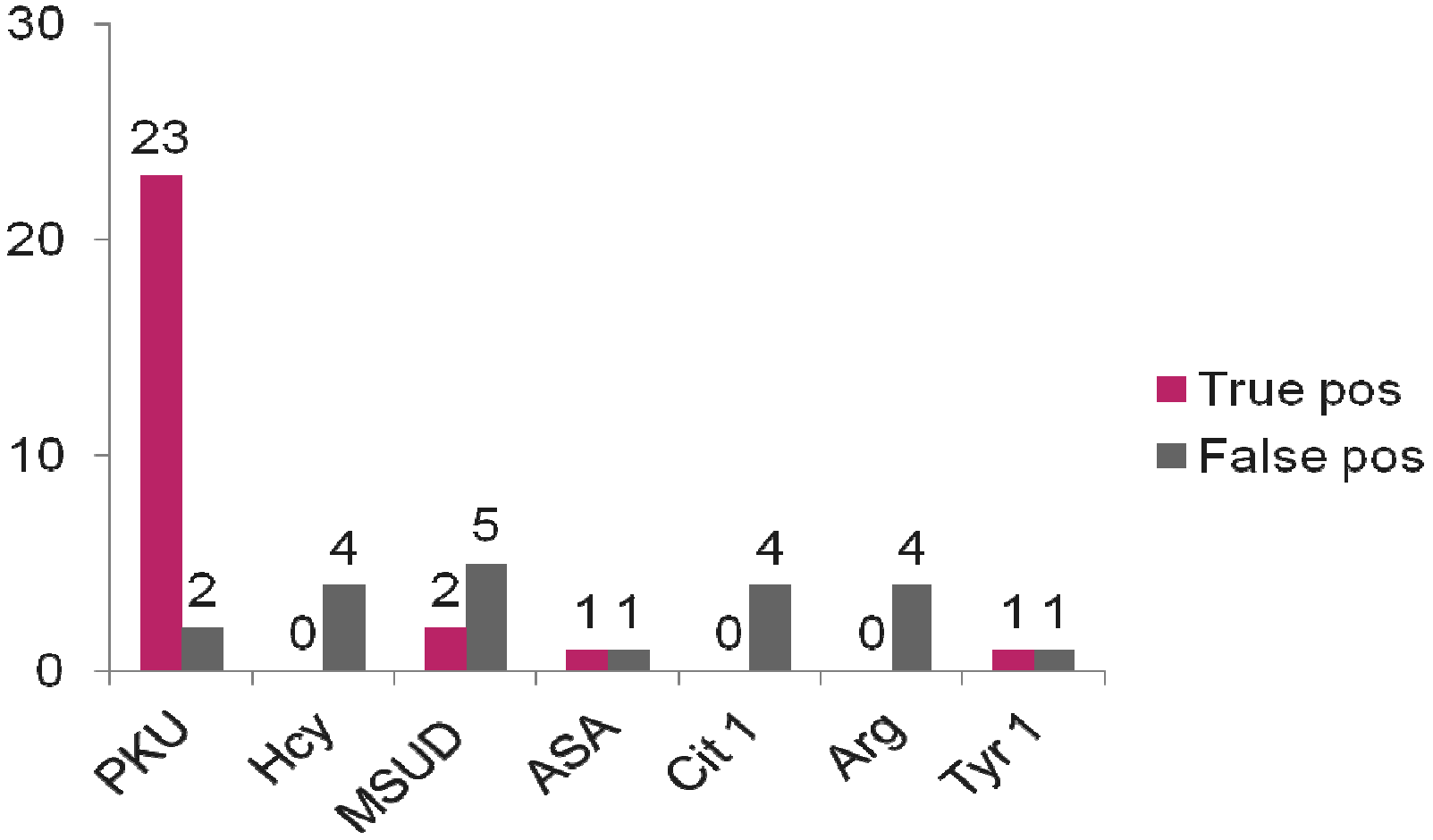
180 000 newborns

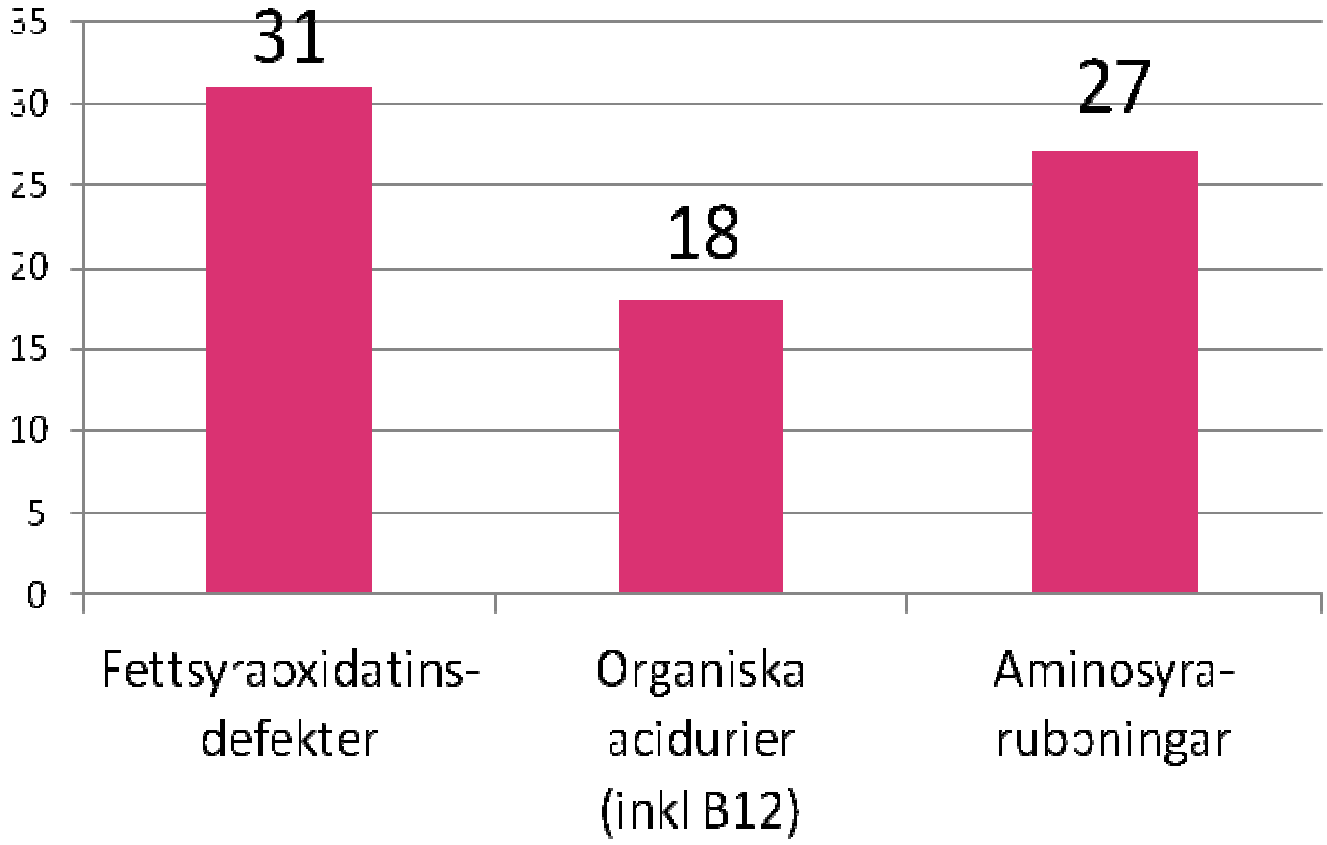


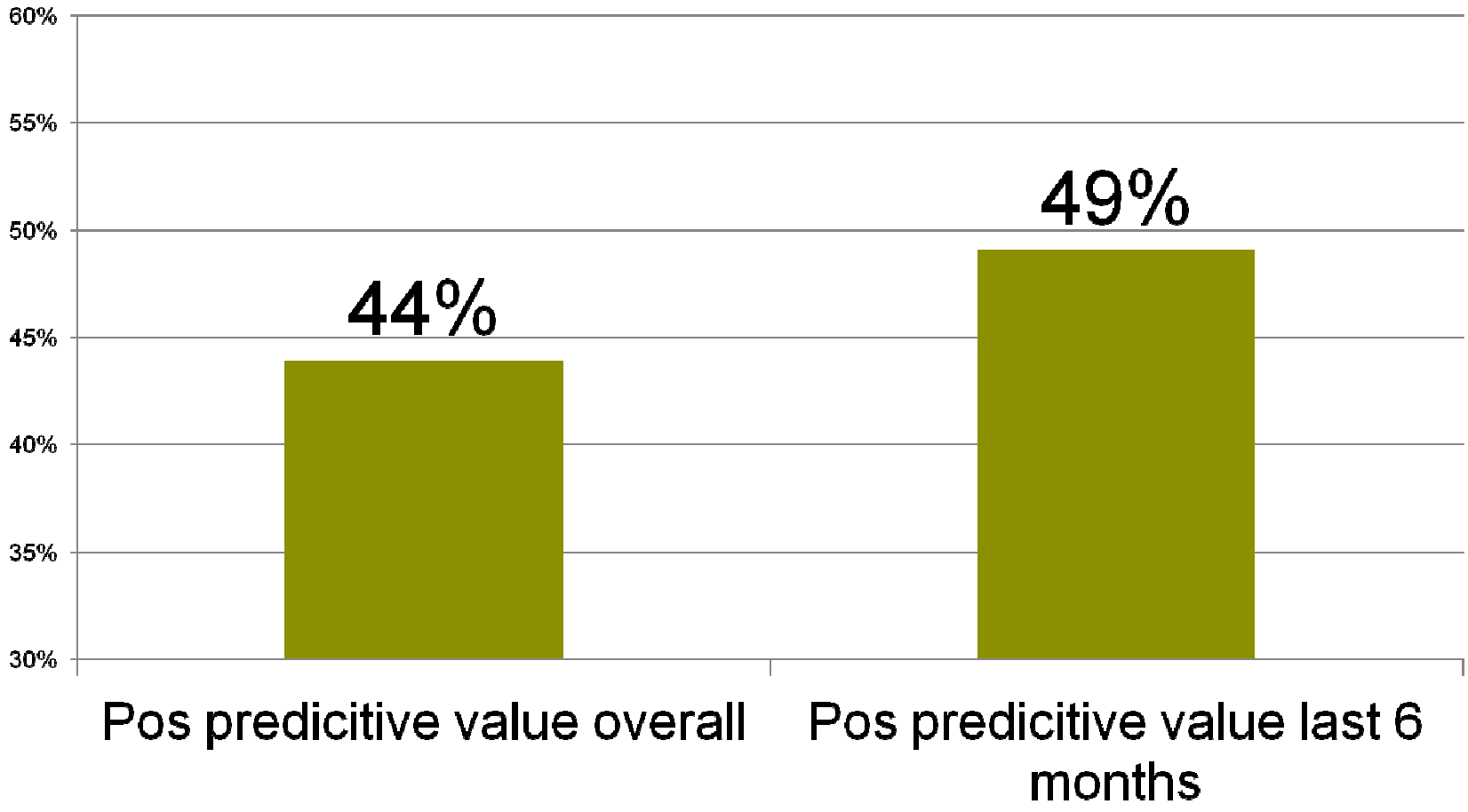
Organic acidurias



Aminoacids


30
20
10
0
23
2
0
4
2
5
1 1
0
4
0
4
1 1
PKU
Hcy
MSUD
ASA
Cit 1
Arg
Tyr 1
True pos
False pos


35
30
25
20
15
10
5
0
31
18
27
Fettsyraoxidatins-defekter
Organiska acidurier (inkl B12)
Aminosyra-rubbningar


60%
55%
50%
45%
40%
35%
30%
44%
49%
Pos predicitive value overall
Pos predicitive value last 6 months

Future

Cystic fibrosis

Immunodeficiencies

Marianne Söderkvist
Susanne Jonsson
Katarina Karlsson
Jenny Ljusberg
Tsige Gulbet
Elin Arvesen
Ann-Chatrine Rosengren
Anna Benedictsson
Ann-Mari Jägstedt

Martin Engvall
Rolf Zetterström
Karin Naess
Elianne Sardh
Anna Wredenberg
Anna Nordenström
Anna Wedell
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