

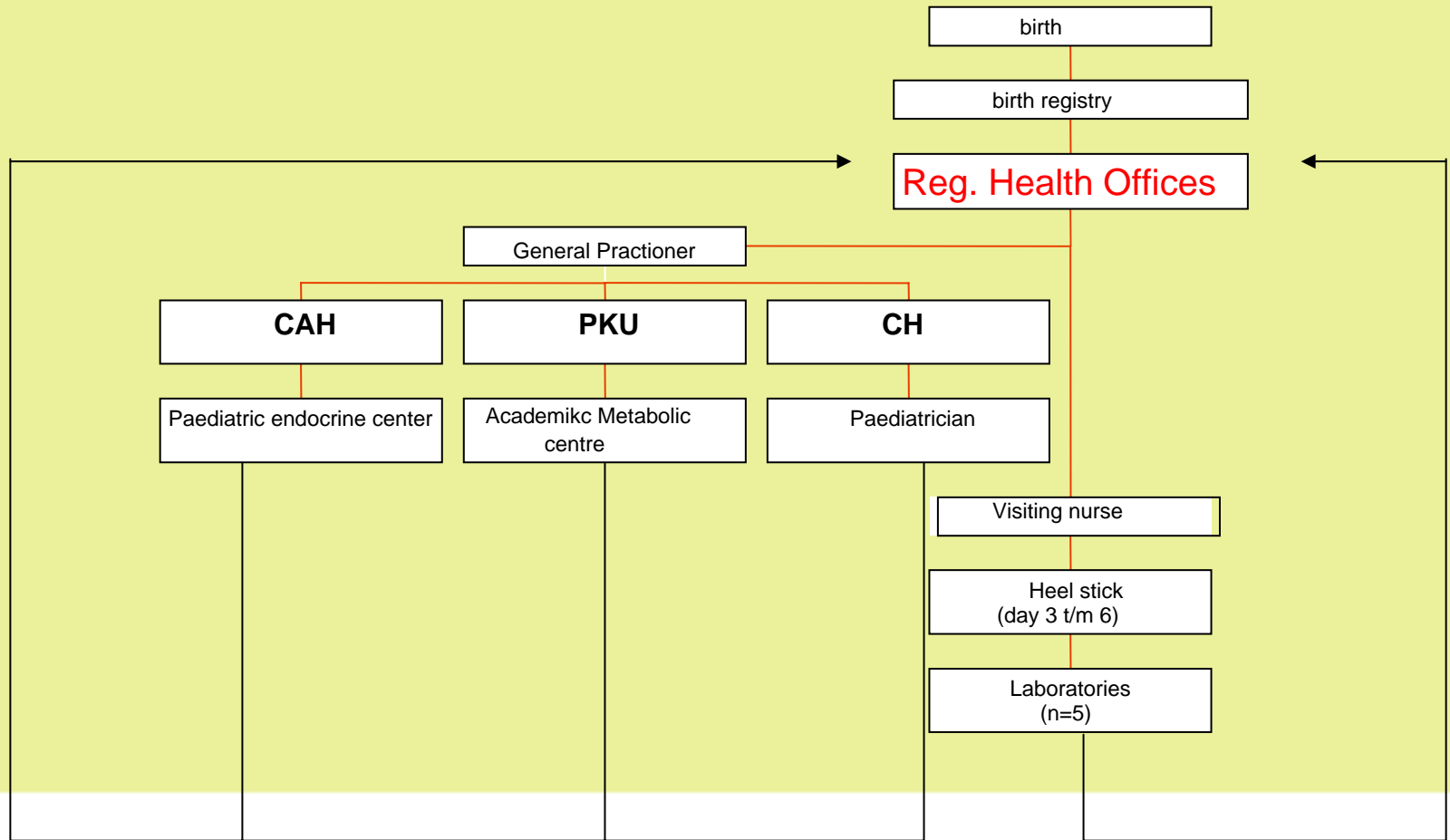


Short term and long term follow up of the neonatal screening program in The Netherlands

J.Gerard Loeber (RIVM) , Paul H.Verkerk (TNO)

Newborn Screening and Genetic Testing Symposium
Minneapolis, May 7-10, 2007

Organisation of the neonatal screening program in NL



Referral, diagnostics, follow-up and treatment

- PKU + other metabolic diseases: 8 academic metabolic centres
- CH: every paediatrician (around 200)
- CAH: 8 academic endocrine centres plus larger regional centres
- SCD: 8 academic hematologic centres

Registration of clinical data

- Each paediatrician reports to TNO all referred children plus clinical findings and follow-up
- TNO assembles annual reports
- Metabolic, endocrine and hematologic paediatricians have formed advisory committees to set up and discuss treatment protocols

Test- and program sensitivity

Coverage	Test sensitivity	Laboratory errors	Follow-up	Sensitivity diagnostic process	Program sensitivity
100%	100%	0%	100%	100%	100%
90%	100%	0%	100%	100%	90%
90%	90%	0%	100%	100%	81%

Validity of screening program in 2005

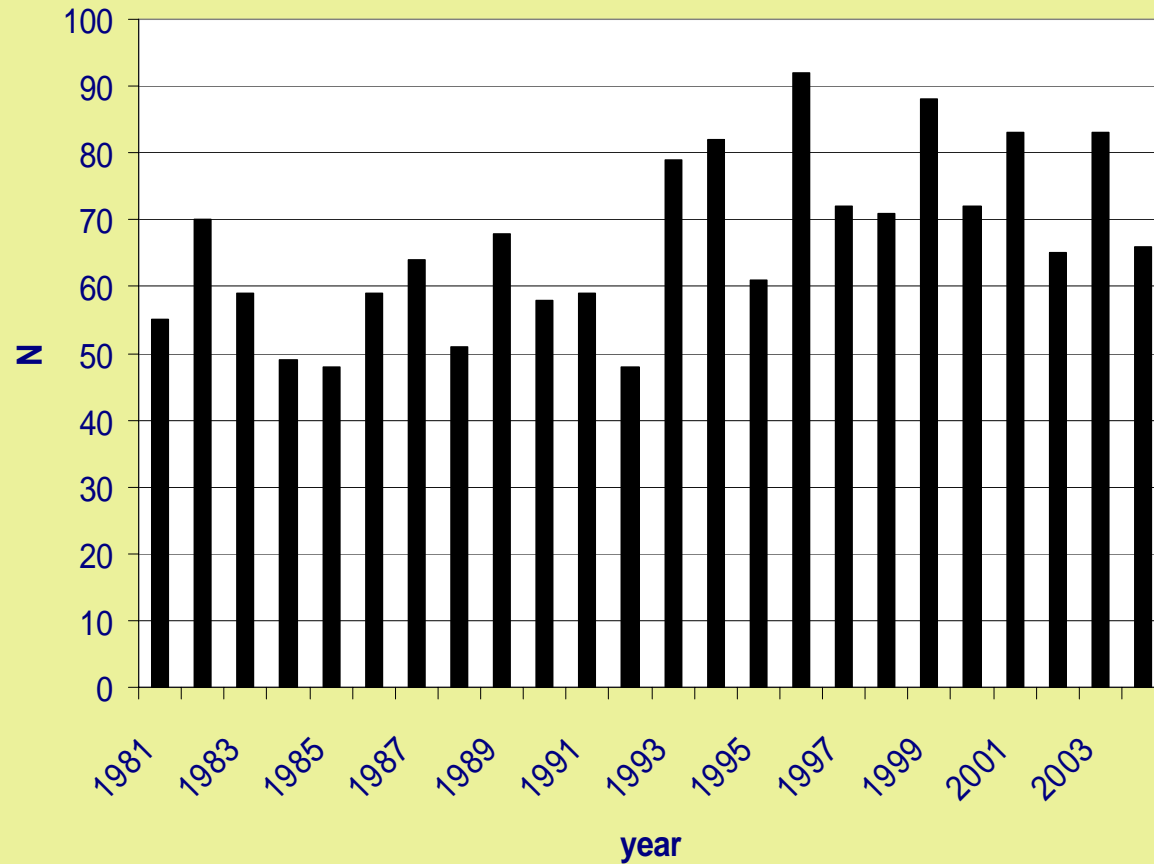
	PKU	CH	CAH
sensitivity	100%	100%	100%
specificity	99,9%	99,8%	99,9%
positive predictive value	55%	22%	19%

Changes in CH-screening procedure

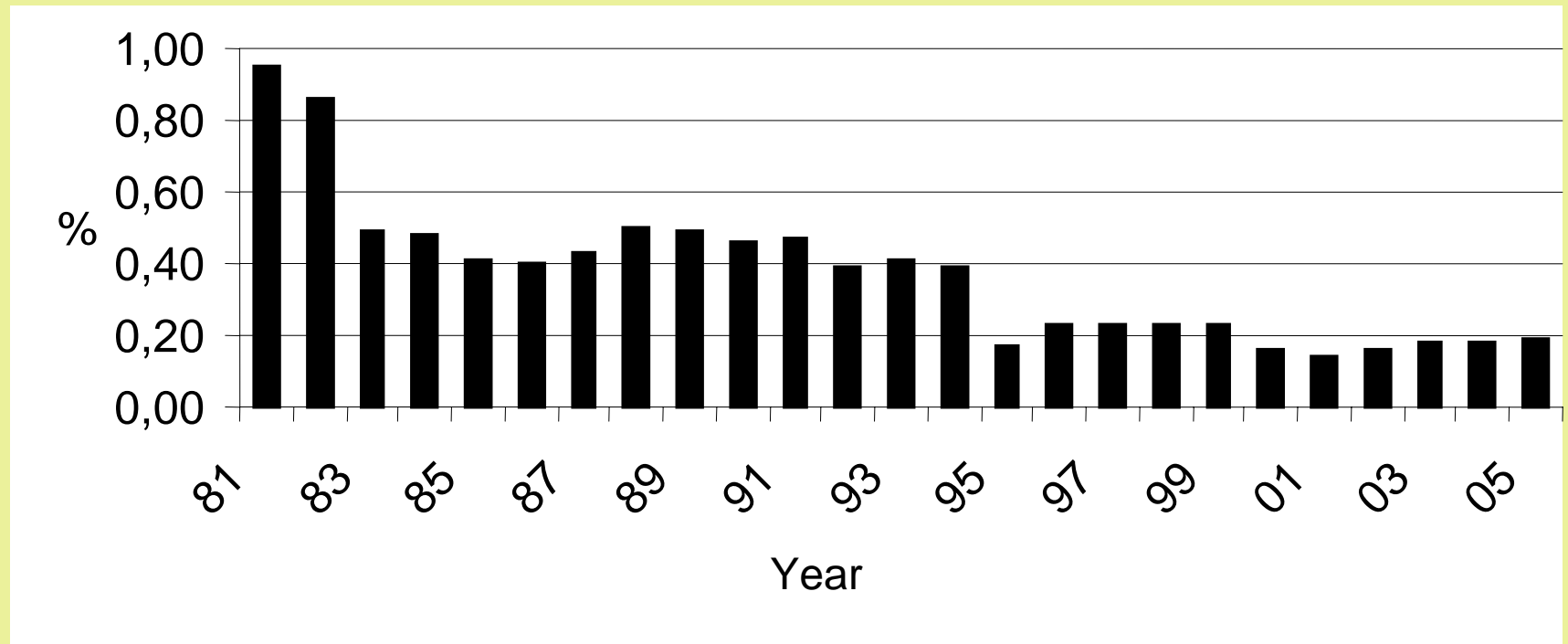
- 1982: prematures only TSH
- 1993: TSH-cut off value from 14 mIU/l blood to 10 mIU/l blood
- 1995: TBG added
- 1996: procedure for older (\geq 60 days) children
- 2005: TSH- cut off value from 10 mIU/l blood to 9 mIU/l blood

Remark: by these modifications both sensitivity and specificity have increased

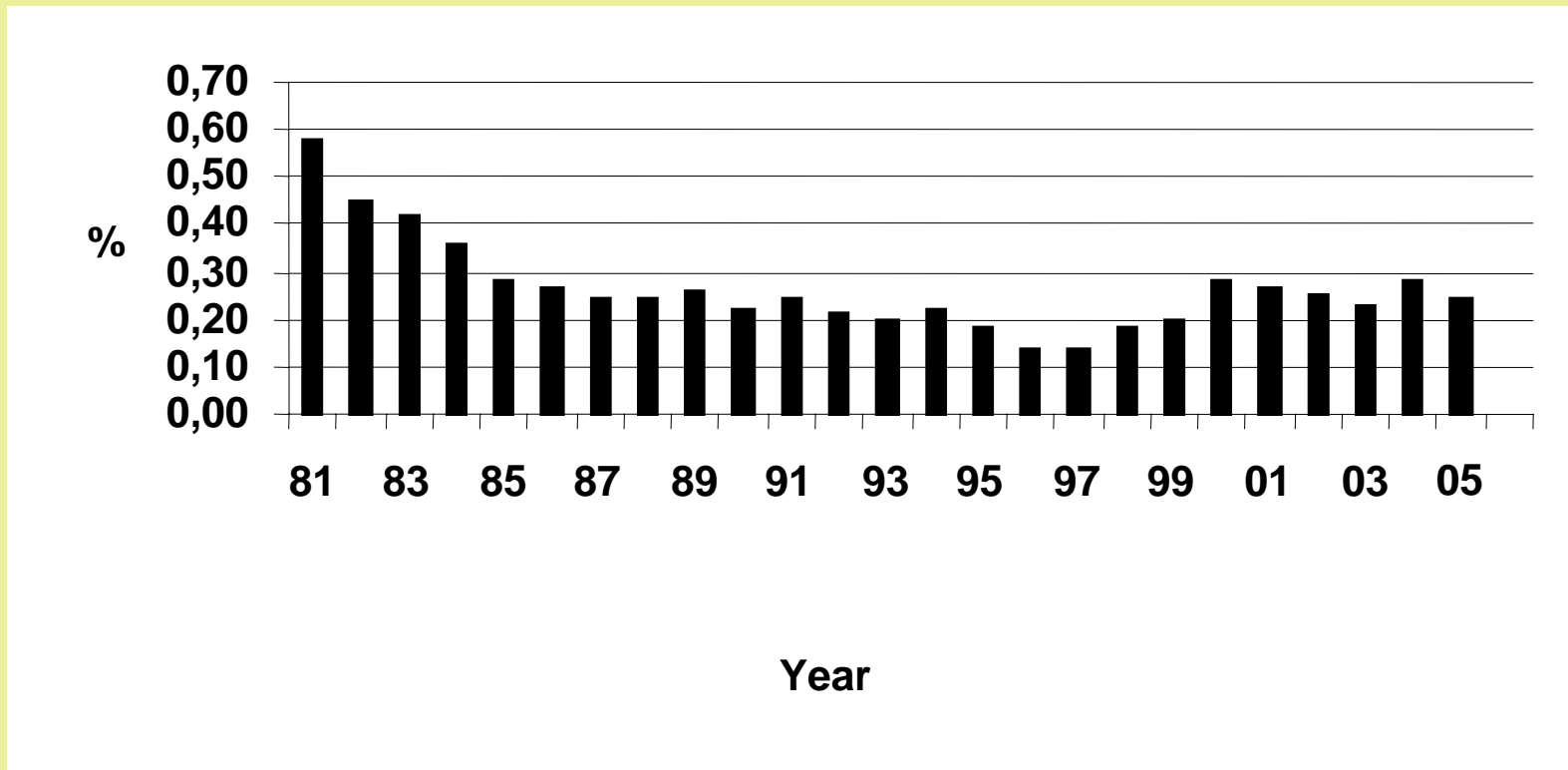
Detected CH patients 1981-2005: n=82



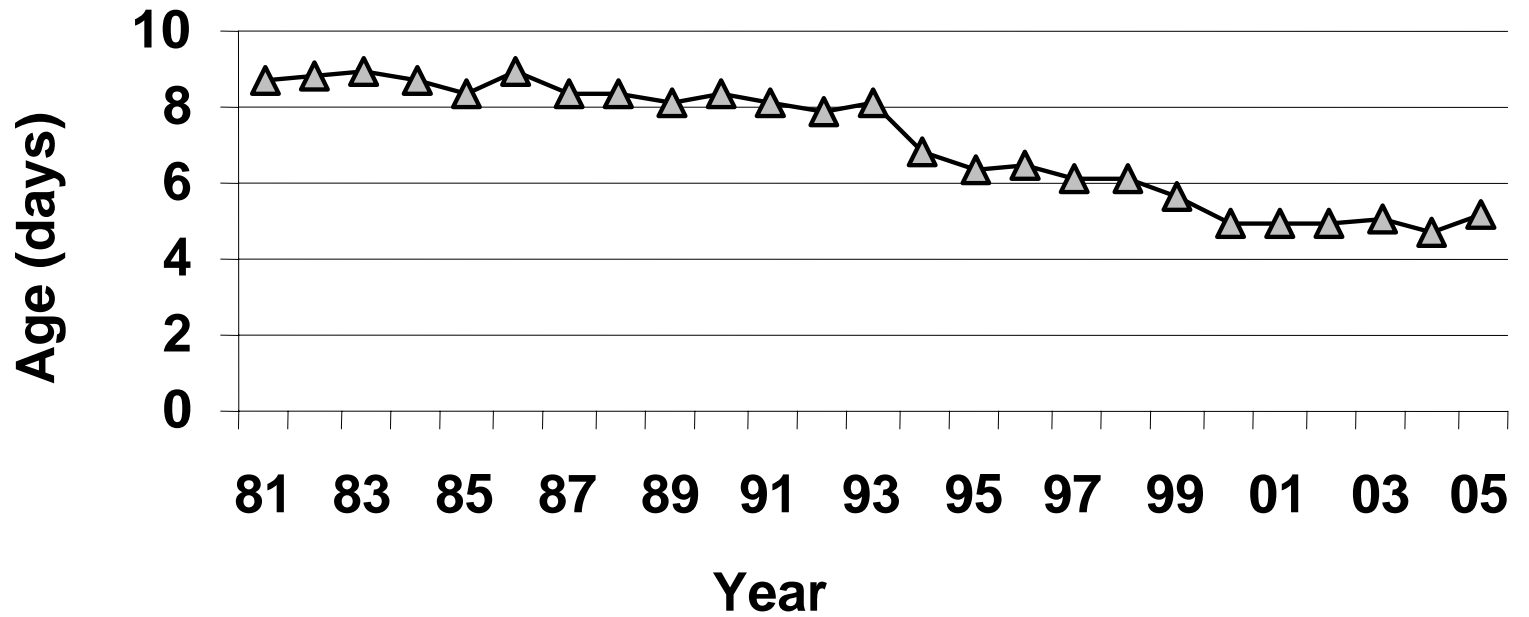
Infants referred to paediatrician for CH-diagnostics



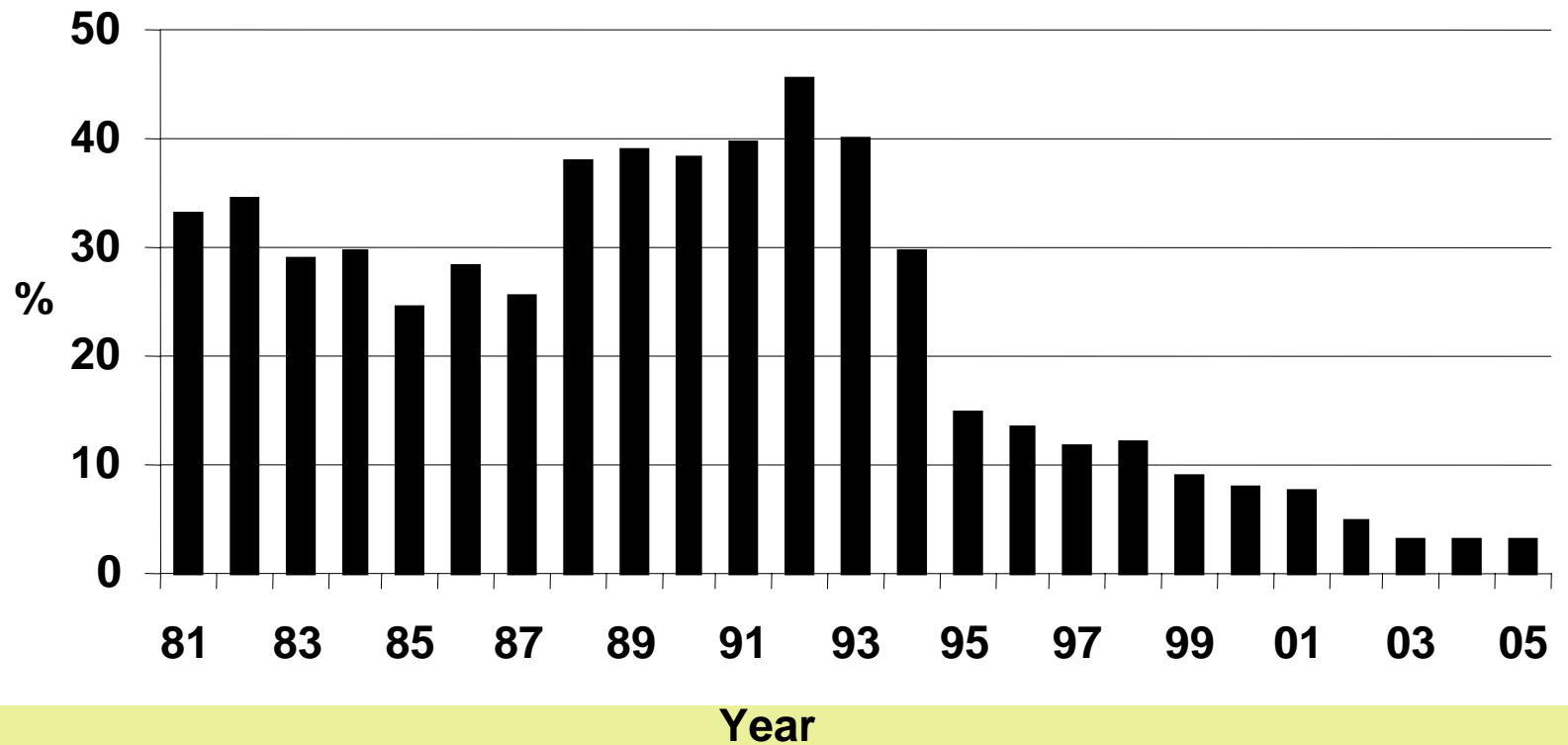
Non-participation



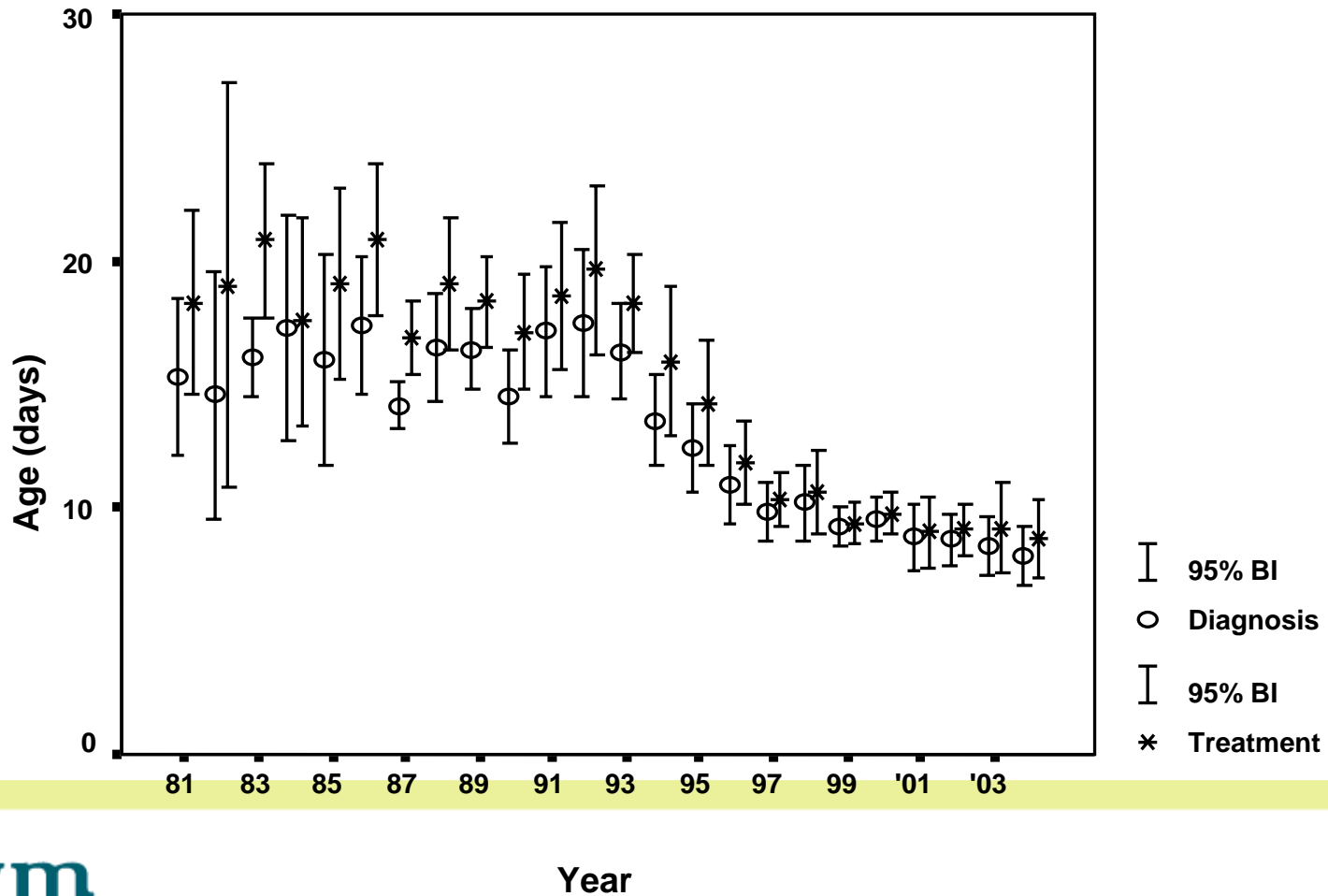
Mean age at first specimen



Interval between report to GP and referral to paediatrician for CH-screening program



Mean age at referral for CH, 1981-2005



Conclusions

- Virtually all patients are detected.
- Virtually all patients are treated early.
- Long term follow-up is essential for monitoring the quality of the entire program.