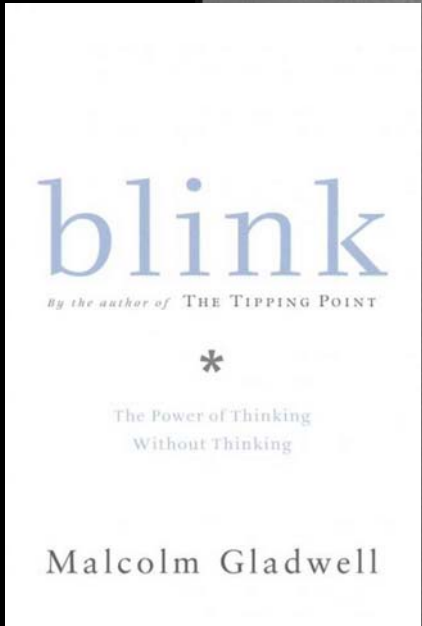


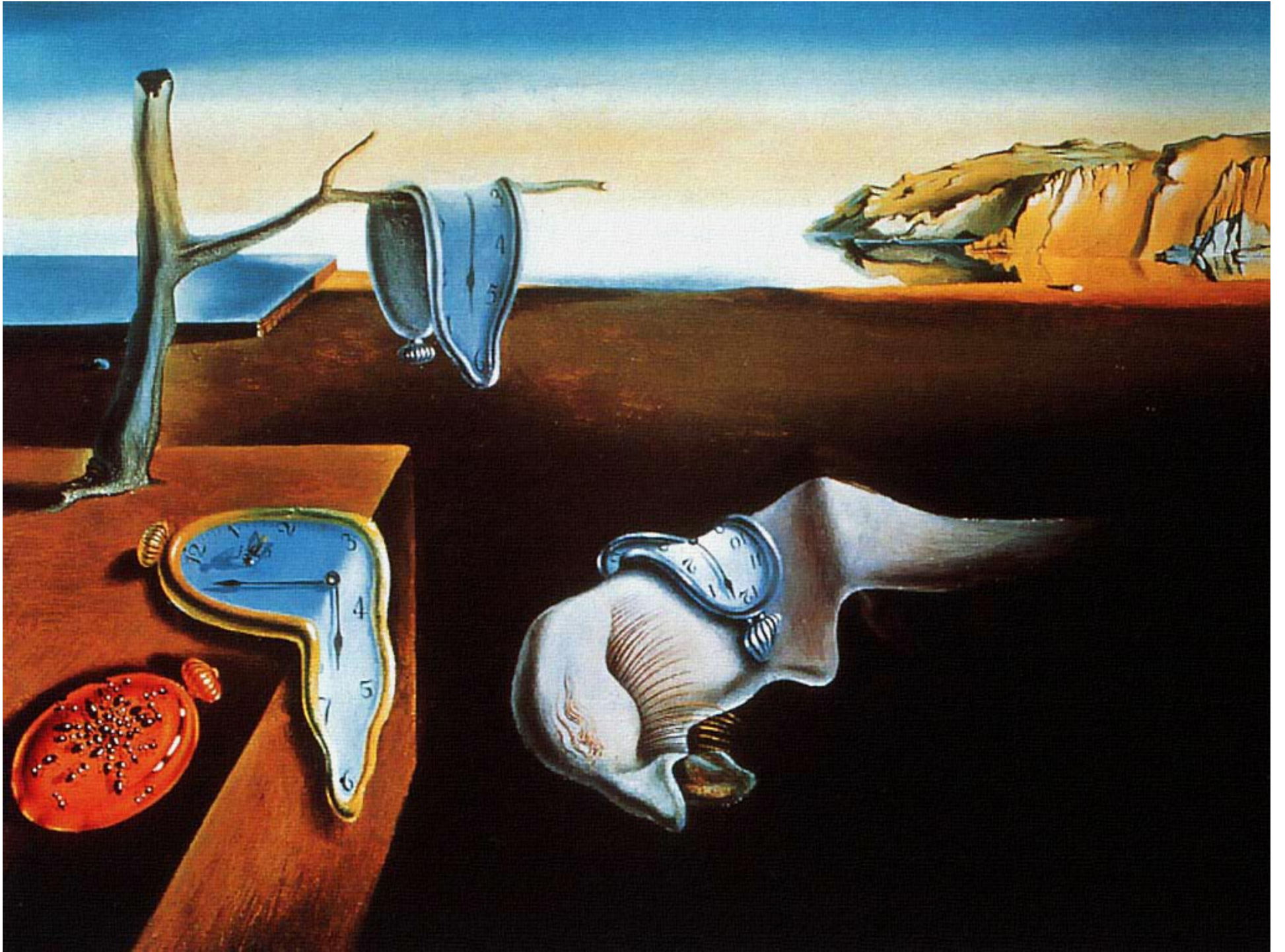


The Art (and Science) of Newborn Screening

John D. Thompson, PhD MPH MPA
Washington State NBS Program







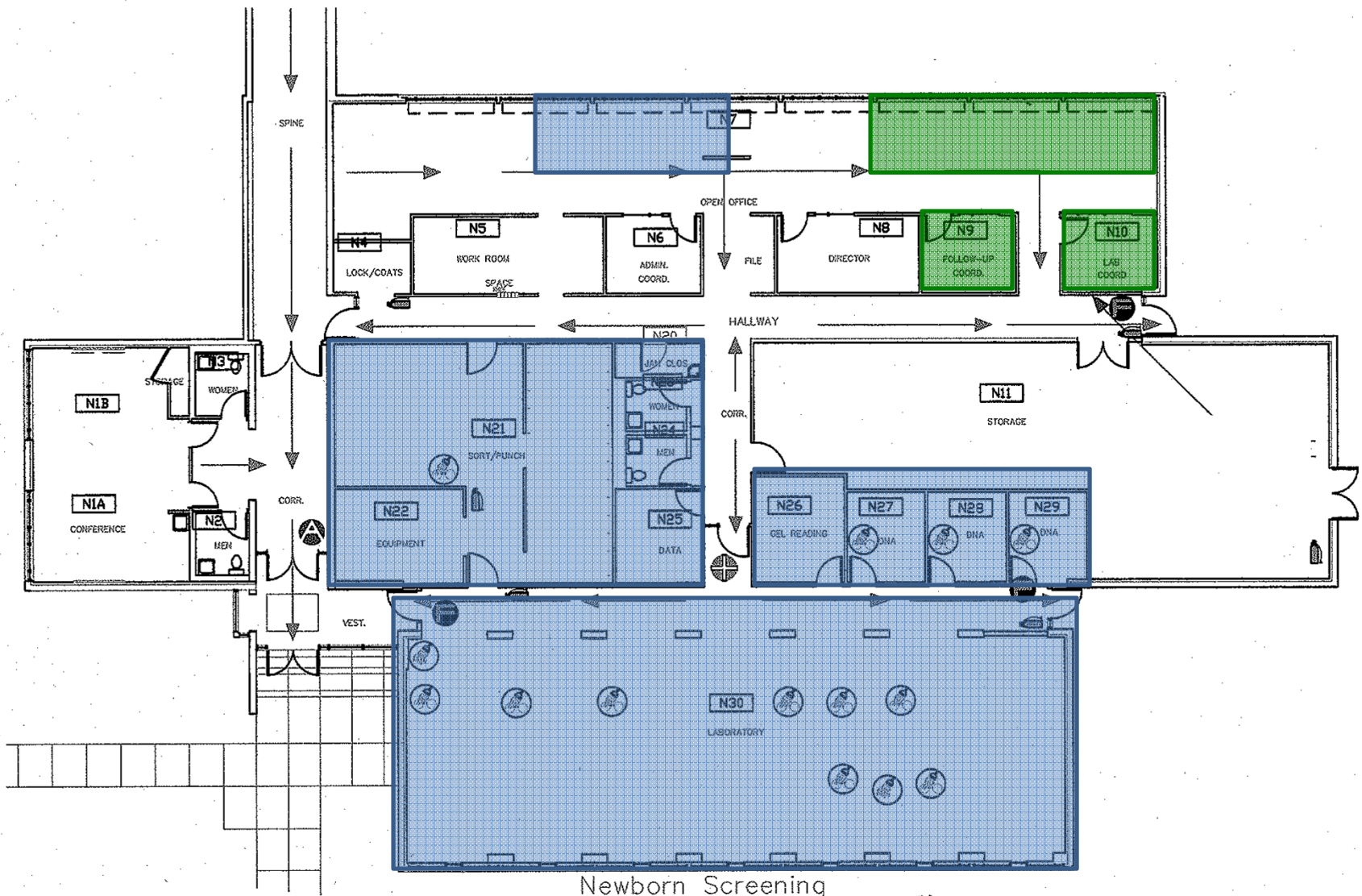


www.treesidefilms.com



www.guardian.co.uk





Newborn Screening



PKU
CH
CAH
HB

200

2



PKU
CH
CAH
HB
BIO
GALT
MCADD
MSUD
HCYS
CF

200

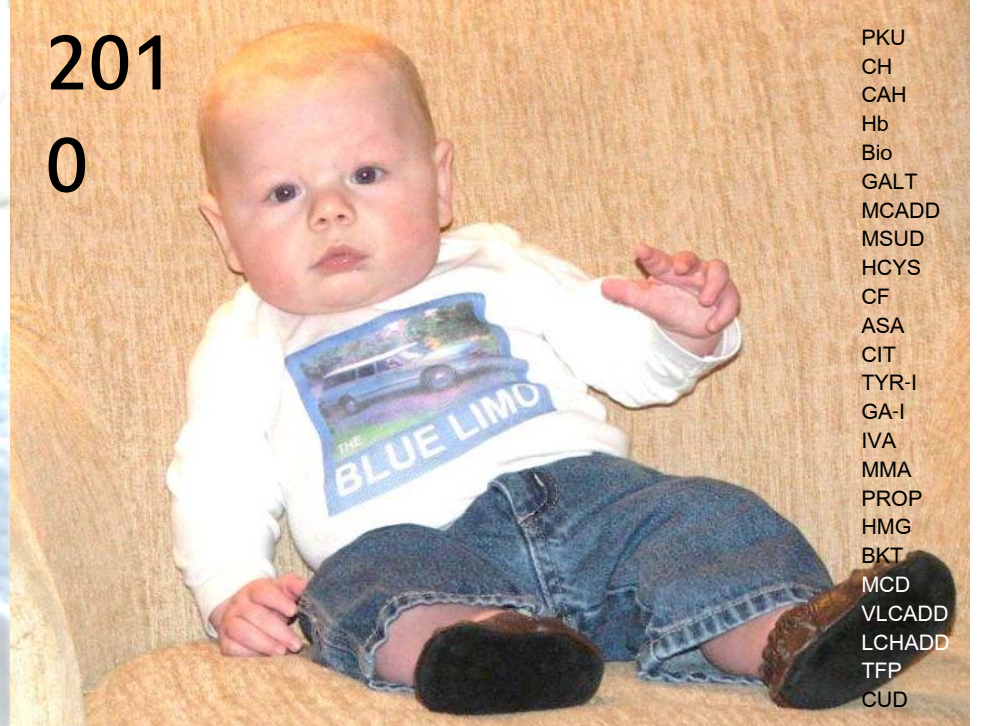
6



PKU
CH
CAH
HB
BIO
GALT
MCADD
MSUD
HCYS
CF

200

8

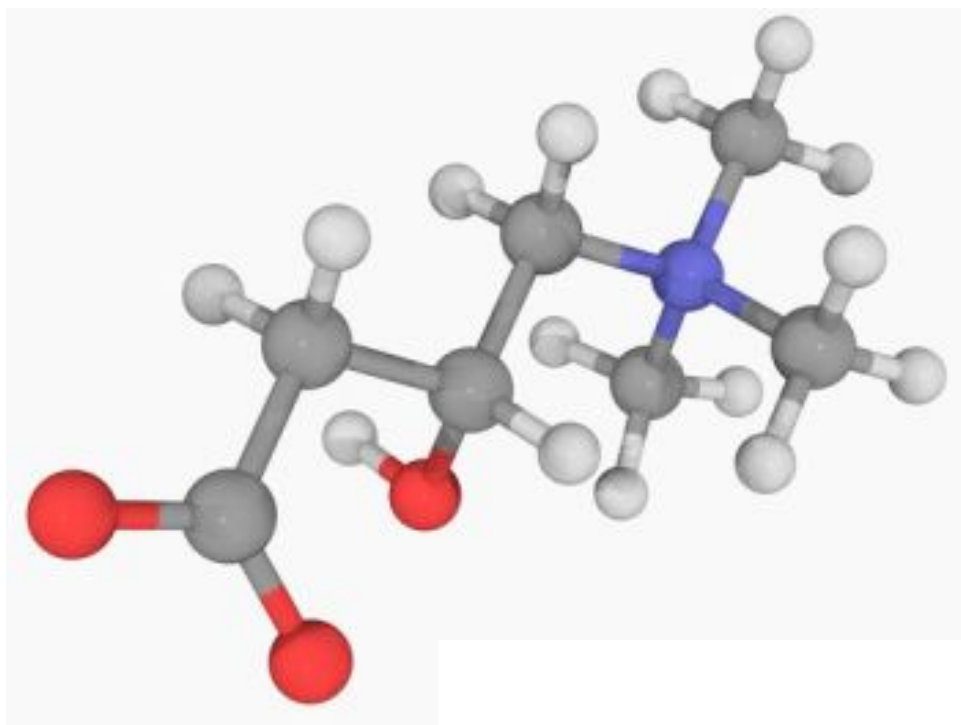


PKU
CH
CAH
Hb
Bio
GALT
MCADD
MSUD
HCYS
CF
ASA
CIT
TYR-I
GA-I
IVA
MMA
PROP
HMG
BKT
MCD
VLCADD
LCHADD
TFP
CUD

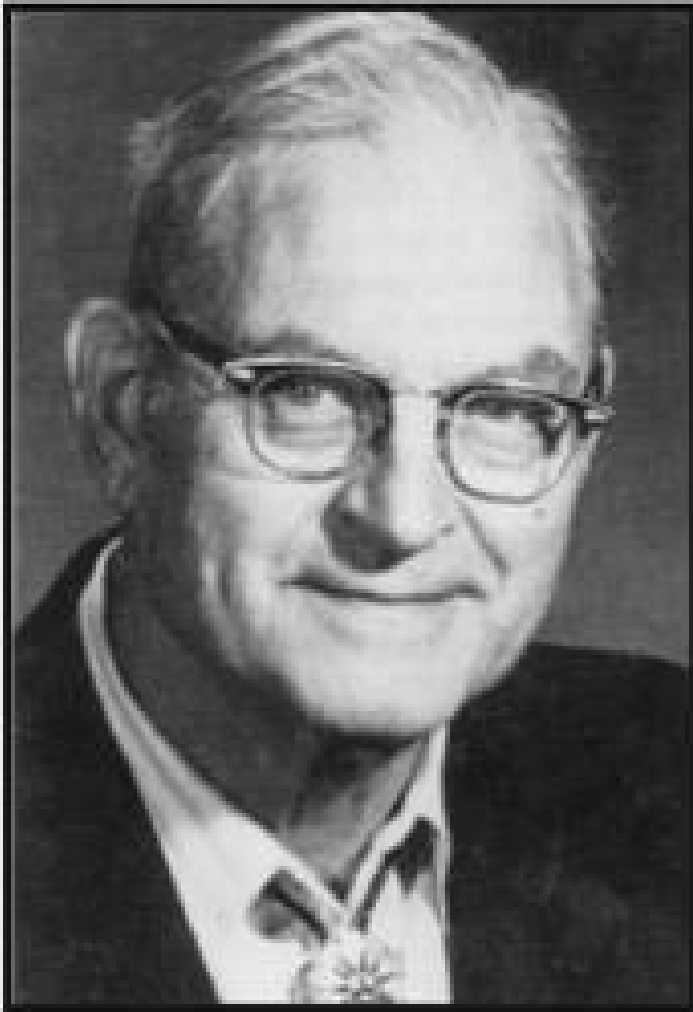
201

0











Art of NBS: Lessons Learned I

- With high-volume experience comes an intuition for follow-up
 - Investigate gut feelings: often they are right
- “Once something is abnormal, normal isn’t always normal” – Sheila Weiss
 - Exercise care in interpreting subsequent screens’ results after a previous specimen was abnormal
- Be creative when you run into dead ends
 - Each case is a puzzle to solve
 - Use all resources available, including colleagues

Art of NBS: Lessons Learned II

- Lab and follow-up can create good synergy working together
 - Look for ways to collaborate and share information, especially when challenges arise
- Pay attention to what the data say
 - Don't be lulled by the high-volume, repetitive nature of follow-up; look for patterns and exceptions
- Regularly evaluate cutoffs and protocols
 - Work with clinical specialists to work out kinks

Art of NBS: Lessons Learned III

- Don't reinvent the wheel
 - See what has been published
 - Ask: a lot has never been published
- Leverage the experience of outside programs
 - Chances are someone else is facing the same problem
 - Power in numbers

Final Thoughts

- Be courteous
- Ask questions!
 - what do/don't we know?
 - what would be important to find out?
 - how are we doing and what could we do better?
- Data are our friends – get acquainted, try to understand them and question them if they seem wrong
- Don't fly on autopilot!
- Where can we get the biggest bang for our buck?