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Newborn Screening Status in South Africa

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What is Newborn Screening?

- Population screening for selected inherited (and other) diseases shortly after birth
- The selected diseases
  - Are serious (Mental disability, Chronic organ failure, Death)
  - No/limited clinical lead time
  - Accurate laboratory (or other) test for diagnosis is available
  - Treatment is available
  - No identifiable risk factors (usually autosomal recessive)
- Comprehensive system
  - Testing, counselling, confirmation, referral, treatment

Global and National Perspective

	Global	SA
Total births (per year)	134M	1M
Predicted incidence of NBS conditions	1:2000	1:2000
Total NBS affected births (per day)	184	1.4
Total screened (per year)	50M	4K
% of all births screened	37%	0.4%
Babies saved through NBS per year	24K	2
Not saved	43K	498

South Africa vs other developing countries

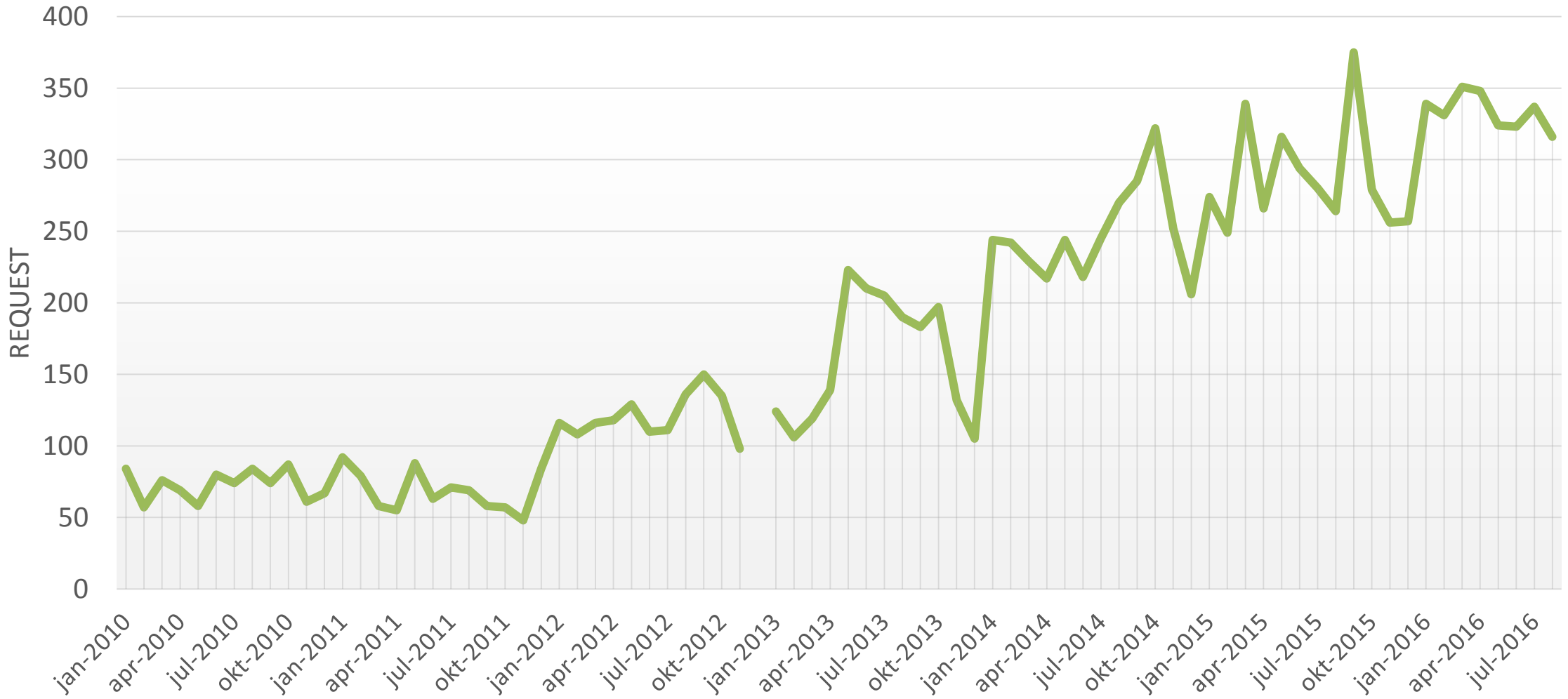
Country	GDP / Capita in 2010 (USD)	% of population screened
South Africa	7,280	< 1%
Egypt	2,591	95%
Iran	5,449	85%
China	4,382	40%
Malaysia	8,519	> 95%
Philippines	2,132	30%
Brazil	10,978	>80%

What does the available program involve?

- 22 Primary conditions
- Fee for service
- National footprint
- ¼ of all private Paeds have requested
- Cost: R 1,200
- Partially reimbursed by Discovery Health
- First advisory board meeting
- Guidelines for diagnosis and treatment

Amino Acid Disorders	
1	Citrullinemia, Type I
2	Classic Phenylketonuria
3	Homocystinuria
4	Maple Syrup Urine Disease
5	Tyrosinemia, Type I
Organic acid disorders	
6	3-Hydroxy-3-Methyglutaric Aciduria
7	3-Methylcrotonyl-CoA Carboxylase Deficiency
8	Glutaric Acidemia Type I
9	Holocarboxylase Synthase Deficiency
10	Isovaleric Acidemia
11	Methylmalonic Acidemia (Cobalamin disorders)
12	Methylmalonic Acidemia (methylmalonyl-CoA mutase)
13	Propionic Acidemia
14	ß-Ketothiolase Deficiency
Disorders of fatty acid oxidation	
15	Carnitine Uptake Defect/Carnitine Transport Defect
16	Medium-chain Acyl-CoA Dehydrogenase Deficiency
17	Very Long-chain Acyl-CoA Dehydrogenase Deficiency
Disorders of carbohydrate metabolism	
18	Classic Galactosemia
Endocrine disorders	
19	Congenital adrenal hyperplasia
20	Primary Congenital Hypothyroidism
Other Disorders	
21	Biotinidase Deficiency
22	Cystic Fibrosis

Requests for NBS in South Africa



Most Common NBS conditions in SA

Condition	NBS Cases (confirmed)	Metabolic lab	SA Incidence	Incidence ref
PKU	1	4.1%	1:100 000 (Black) <1:10 000 (White)	Ann Hum Genet (2008) 72,65-71 J Med Genet (1990) 27,760-779
Tyrosinemia Type I	0	5.4%		
Glutaric acidemia Type I	1	3.8%	1:5 000 (Black)	Mol Gen Metab (2010) 178-182
Isovaleric acidemia	2	4.5%		
Methylmalonic acidemia	3	5.7%		
Propionic acidemia	2	6.3%		
Galactosemia	3	5.5%	1:14 000 (Black)	BMC pediatrics (2002),2:7
Congenital Hypothyroidism	4		1:4 000 (White)	J Med Genet (1990) 27,760-779
Cystic Fibrosis	NA		1:2 000 (White) 1:12 000 (Mixed) 1:32 000 (black)	South African Cystic Fibrosis Trust Website
Biotinidase deficiency	2	1.7%		
MSUD	1	1.6%		

Big names – easy treatment

Condition	Discipline	Management	Treatment	Care complexity	Care Cost
PKU	Neurology	Dietician	Diet	High	High
Tyrosinemia Type I	Gastroenterology	Gastroenterology	NTBC, Liver transplant	Very high	Very high
Glutaric acidemia Type I	Neurology	Dietician, General paediatrician	Low lysine diet, Carnitine, Intermittent glucose	Low-Medium	Low
Isovaleric acidemia	Neurology	Dietician, General paediatrician	Low protein diet, Glycine, Carnitine	Low	Low
Methylmalonic acidemia	Neurology	Dietician, General paediatrician	Low protein diet, Vit B12, Carnitine	Low	Low
Propionic acidemia	Neurology	Dietician, General paediatrician, Specialist support	Low protein diet, Other	Medium	Low
Galactosemia	Gastroenterology	Dietician, General paediatrician	Avoid galactose	Low	Low
Congenital Hypothyroidism	Endocrinology	Dietician, General paediatrician	Eltroxin	Low	Low
Cystic Fibrosis	Pulmonology	Paediatric pulmonologist		Medium – High	High
Biotinidase deficiency	Neurology	General paediatrician	Biotin	Low	Low
(MCAD)	Neurology	Dietician, General paediatrician	Avoid fasting	Medium	Low

So why are we not screening our children?

- What motivation
  - Economic
  - Human rights
  - Legal
  - Professional
  - Political
- Whose perspective
  - Family
  - Health care provider
  - Medical aids
  - Government

Economics and Human Rights

1. South African constitution
2. UN convention on the rights of the child
3. African charter on the rights and welfare of the child
4. UN convention on the rights of persons with disabilities

Equal before the law
Equal protection and benefit
No discrimination, stigmatization or marginalization
Highest attainable standard of health
Family environment
Happiness love and understanding

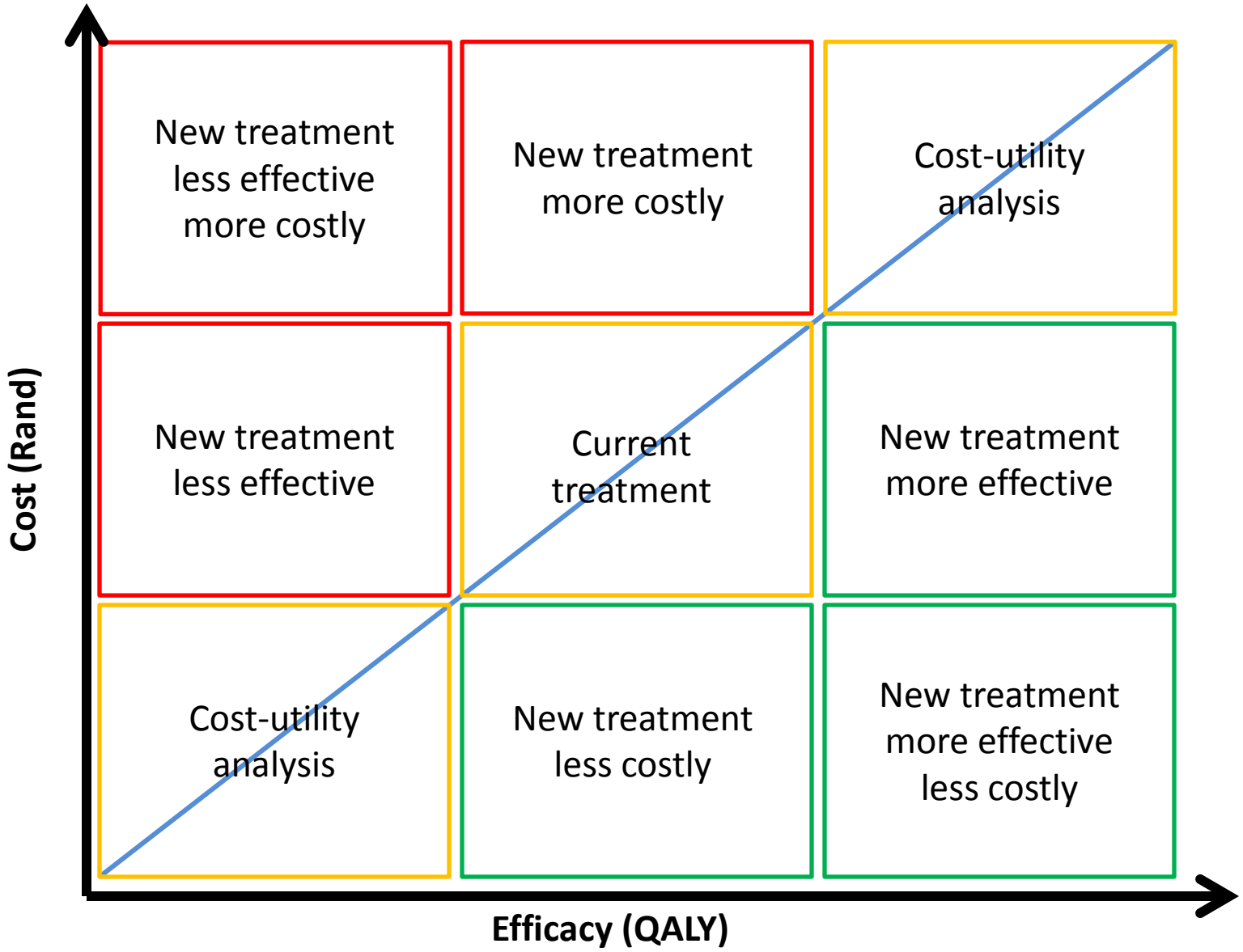
Screen and prevent
vs
Manage complications

Economic analysis

Cost minimization analysis (CMA)
Cost-utility analysis (CUA)

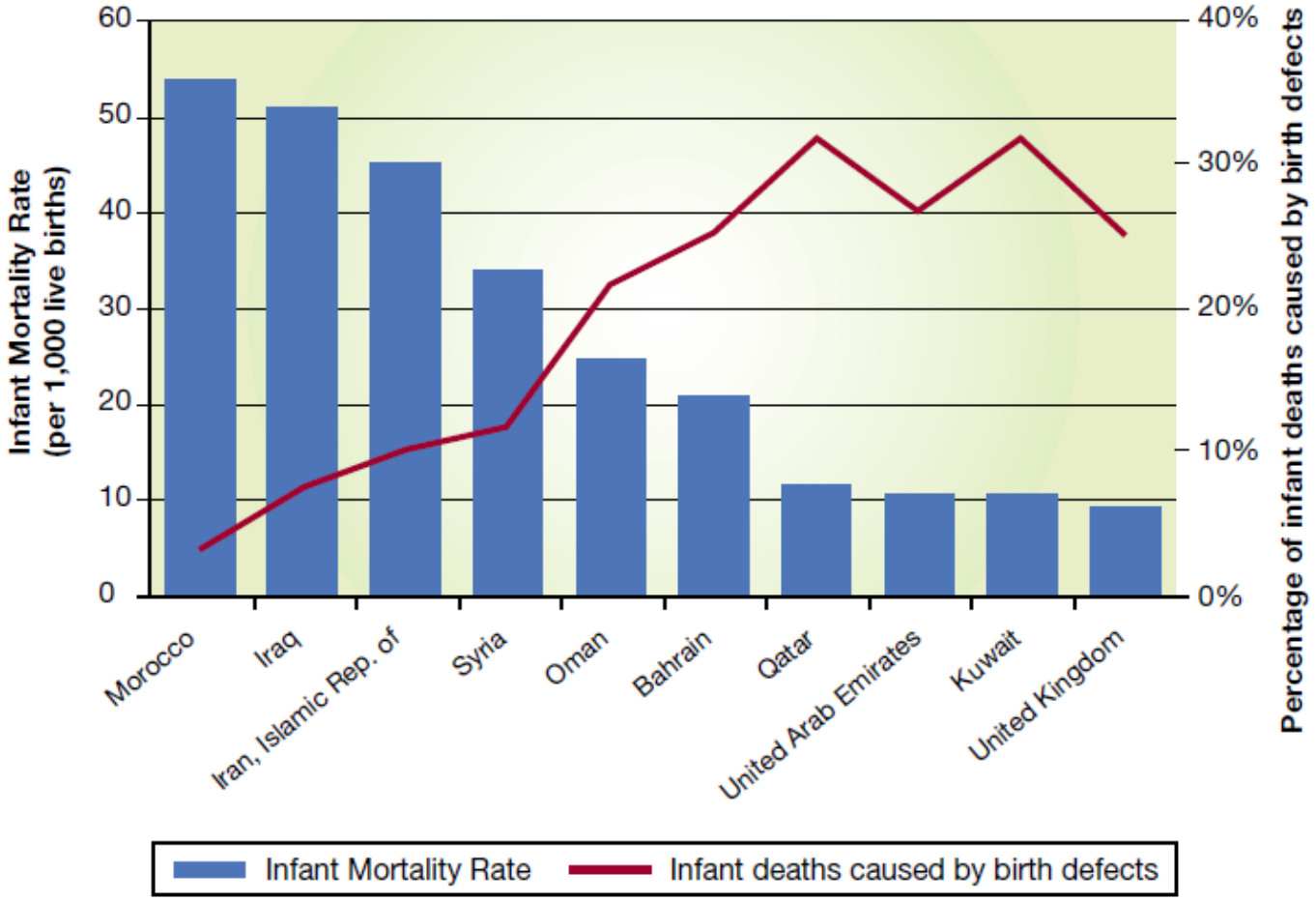
Economic perspective

$$CUA = \frac{Cost}{QALY}$$



Contribution of birth defects to the infant mortality rate

Relationship between Infant Mortality Rate and Percentage of Infant Deaths due to Birth Defects by Country

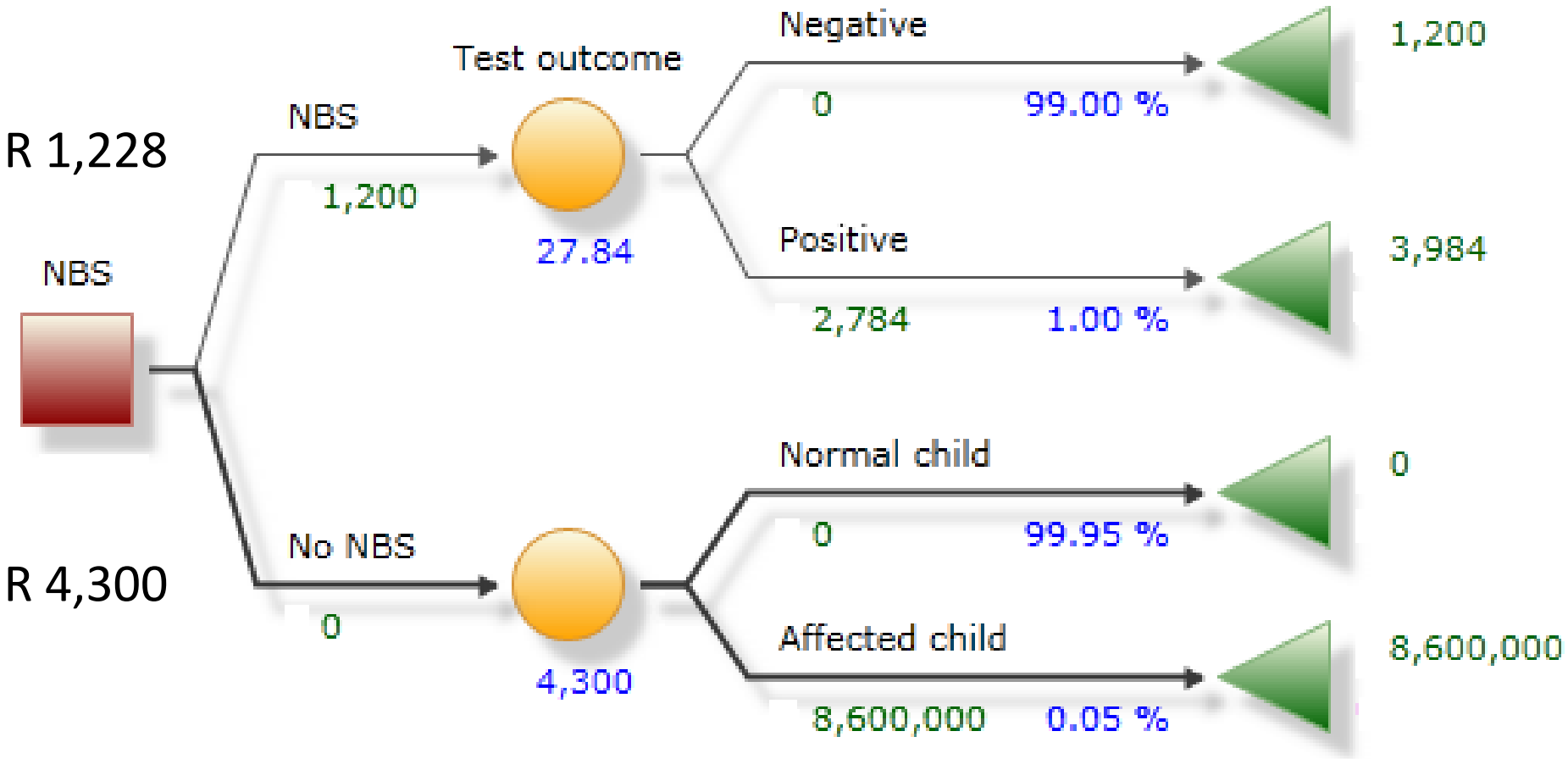


SOURCE: *Adapted from WHO, 1997.*

Ballpark CMA and CUA of NBS

	Private NBS	Private patient	Public NBS	Public patient
Child raising	0	0	0	0
Medicine	0	0	0	0
Cost of screening	1,200	0	780	0
% requiring additional testing	1%	0	1%	0
Ave cost of additional testing	2,784	0	1,810	0
Total screening cost	1,228	0	798	0
Incidence	0.05%	0.05%	0.05%	0.05%
Cost of finding a positive case	2,455,680	0	1,596,192	0
Cost of diagnosis	0	200,000	0	130,000
Life expectancy	70	70	70	70
NPV of special care cost	0	8,400,000	0	1,344,000
Total cost	2,455,680	8,600,000	1,596,192	1,474,000
QALY	1	0.65	1	0.55
Total QALYs	70	45.5	70	38.5
CUA	35,081	189,011	23,771	38,286

Parent / Clinician perspective



Medical scheme perspective

Members (2014)	8,8M	
Main members (2014)	3,9M	
Dependants (2014)	4,9M	
Total contributions (2014)	R 140,2B	
Average per main member (2014)	R 35,754	
Average per main member (Monthly)	R 2,980	
Private health care births (Annual)	166 946	
	NBS	No NBS
Probability of incurring cost	100.00%	0.05%
Cost per incident	R 1,228	R 200,000
Total Cost per main member	R 205M	R 17M

Other (lame) excuses

- Lack of infrastructure
- Lack of expertise
- Lost to follow-up
- NBS diseases only affect white people

Why are we not screening our children?

Lack of political incentive

Lack of willpower / complacency

Ignorance

#MentalDisabilityMustFall

Newborn Screening in South Africa

www.newbornscreening.co.za

https://nextbio.co.za/firstscreen/

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Thank you