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Newborn Bloodspot Screening: The Importance and Impact

Welsh Paediatric Society Autumn Meeting

08/11/24



Sgrinio Smotyn Gwaed
Newydd-anedig Cymru
Newborn Bloodspot
Screening Wales

- What is population based screening & why is it different?
- NBSW history & delivery
- NBSW clinical conditions, prevalence & specifics

10
year
anniversary

Population based Screening: **Pre-requisites**

WHAT

- Important health problems
- Well defined diseases or conditions
- Natural history of the disease is known
- Detectable pre-clinical stage

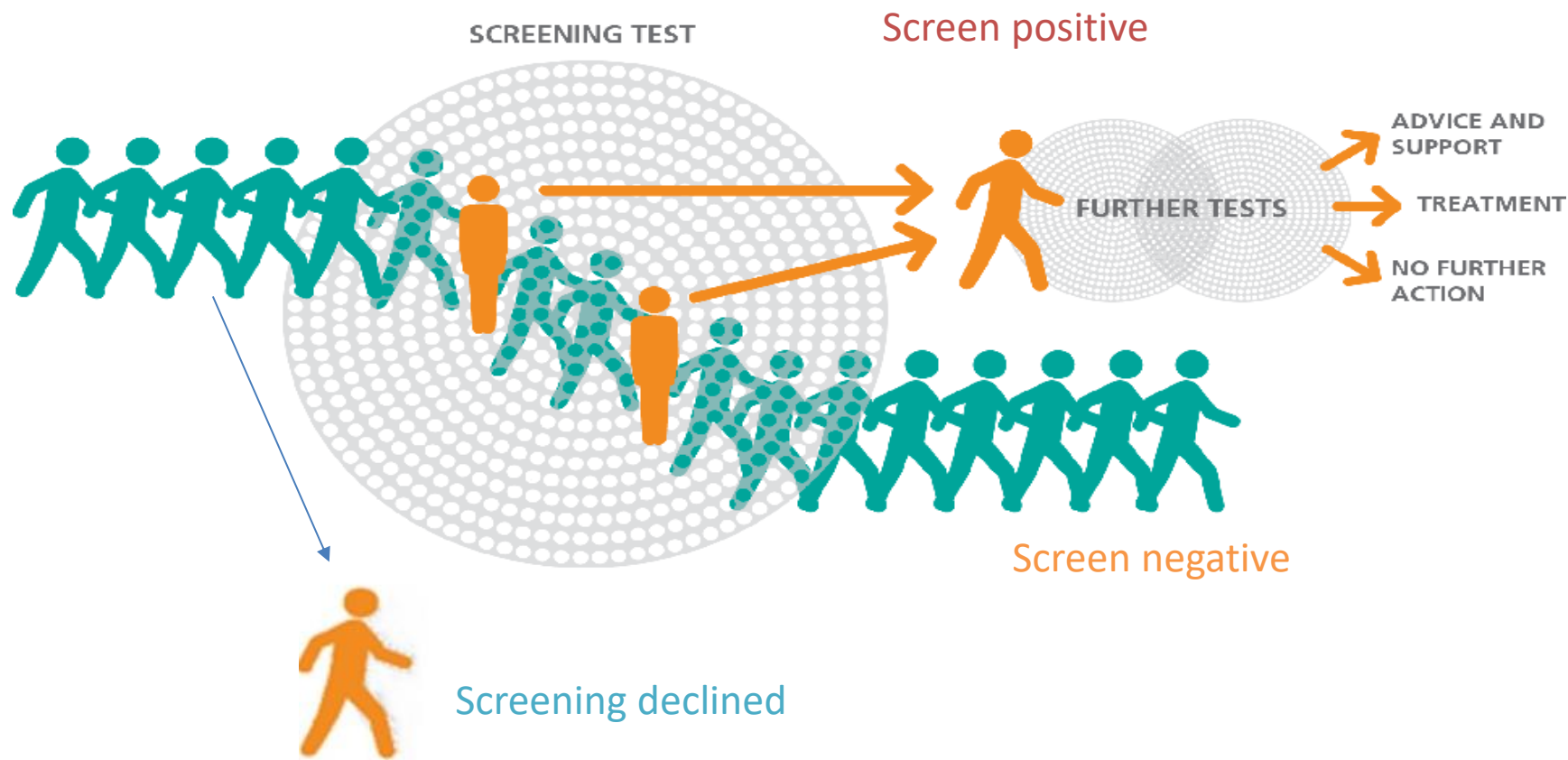
REQUIREMENTS

- Accurate & acceptable test
- Access to treatment & advice for people with the condition
- Does more good than harm to those screened

Population based Screening: **What it is & isn't**

- ✓ Members of a **defined** population
- ✓ People without ... symptoms
- ✓ Personally invited/offered
- ✓ To identify those individuals who may be at increased risk of a disease/condition
- ✓ Enables earlier identification/treatment
- ✓ To **reduce the risk** of disease or its complications
- ✗ A diagnostic service
- ✗ A treatment service – it's not for people with relevant symptoms
- ✗ A requirement – there must be informed choice
- ✗ Opportunistic testing – all the eligible population must be invited

Population based Screening: **The Sieve**



Population based Screening: **Big Differences**

- We make the approach / offer
- Screening programmes aim to do more good than harm at the *population* level
- **Screening is not 100% accurate**
- Inherent irreducible risk of harm
- Screening is a pathway not a test
 - **Screen positive results always lead to diagnostic assessment**

What is Newborn Bloodspot Screening (NBS)?

- Newborn bloodspot screening is when a small sample of blood is taken from the baby's heel on **day 4-6** of life, (counting day of birth as day 0)
- This blood sample is screened for up to 9 rare but serious conditions that respond to early intervention to reduce mortality and/or morbidity
- All babies born or resident in Wales up to the age of 1 are eligible



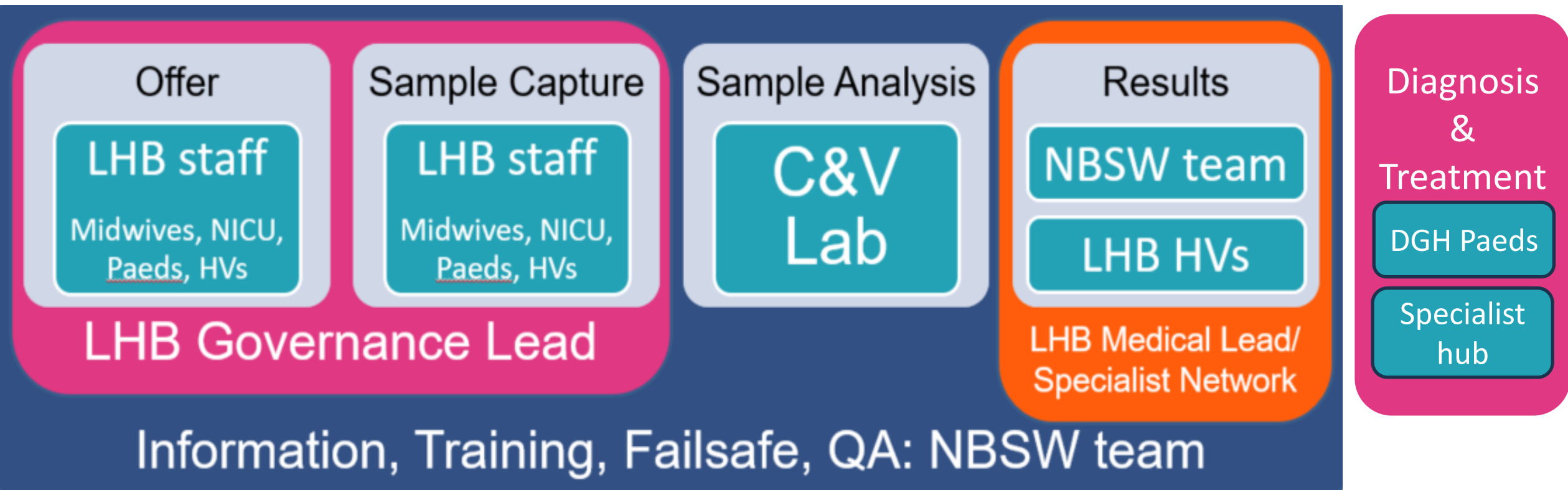
How it all started <2014

In November 2010, the Minister for Health and Social Services asked Public Health Wales to:

- Ensure a sustainable and safe national newborn bloodspot screening programme across Wales
- Introduce screening for MCADD and SCD (UK NSC recommendation)

2011 – 2014: Programme development project

NBSW Programme Delivery



10 years of NBSW: our achievements so far

- Excellent services through partnership delivery
- Screening provided for over **338,000** babies (>95% coverage)
- Successfully introduced screening for 4 additional conditions
- Ensuring consistently high-quality sample capture & timely delivery

Horizon Scanning

- Confirmed/ expected new conditions
 - ✓ Hereditary Tyrosinemia Type 1
 - ? Severe combined immunodeficiency (SCID)
 - ? Spinal Muscular Atrophy
- Genomics
 - As a Screening tool (replace, refine, expand)
 - Treatments enabling new screened for conditions