



Parents' perspective on NBS
for Duchenne Muscular Dystrophy

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Programs

Over 45 years of experience in NBS for DMD

Long history with pilot studies and longer programs (primarily using CK)

Between 1975 and 2011, there have been 17 piloted newborn screening programmes in 11 countries or regions



Parents are in favor of NBS for DMD (since 1997)

Good parenting

Access to appropriate medical care

Access to new treatments

Family planning



Opportunity for good parenting <> 'carefree period'





Carefree period?



“In some ways, I was never at ease about it, but you have yourself persuaded that you need not to worry at all. And this works for a while, just until you notice the next thing; this also goes slow”

“Well, of course I found him different but when they [the doctors, AP] say; don’t worry about it, he will be alright. Then I think; okay, it is fine because they know best, don’t they?”

“You just want clarity, and even though it is the worst clarity, that doesn’t matter that much”

“I would not have been so hard on him when he was three years old, saying ‘we are not going to carry you.’ I would have treated him more caution. You really feel guilty afterwards,..”

“Well, there has been a period that the physiotherapist was busy doing the wrong things. Therefore, it would be better knowing this earlier”

“ I kept on searching and asking to find out what was wrong with my son, for I knew for sure that there was something wrong with him, I just couldn’t get the right answers”



Parents are in favor of NBS for 'Untreatable' conditions

Prospect parents

Parents of Healthy children

Clinicians for their own children

Parents of affected children



Situation in Europe

No programs ongoing

Different opinions and regulations in different countries

Wilson and Jung Criteria.

Treatable versus Actionable

Major delays when you start when a treatment is available
(\leftrightarrow approved, SMA, Translarna)

Harmonization would be something to strive for but should not cause delay

Eurordis Principles

New treatments



Several drugs for DMD in development

Results in younger patients better than in more advanced cases

First drugs (conditional) approved

Setting up NBS when drugs are on the market,
will skip a large cohort of children from opportunities

Catch 22

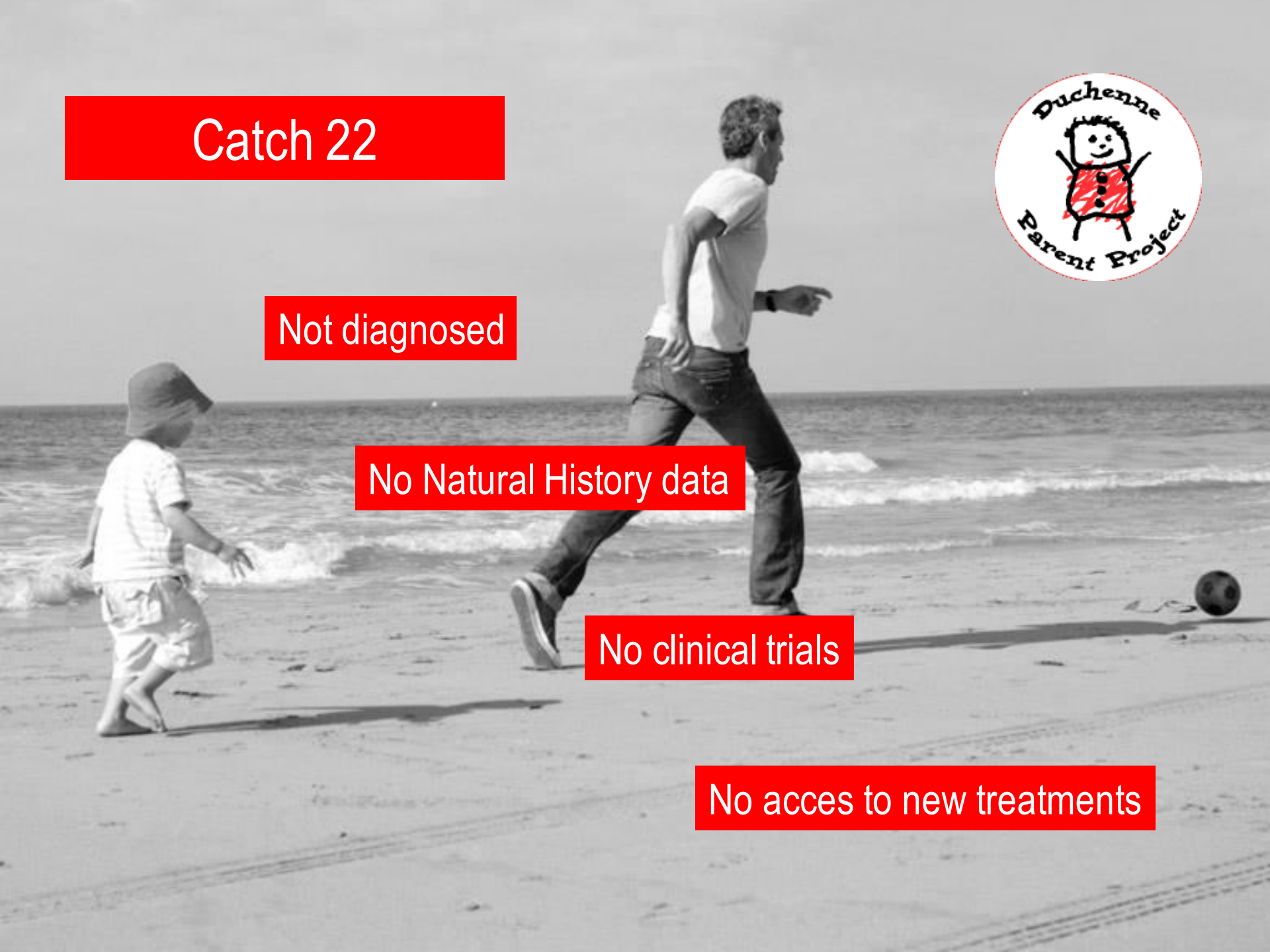


Not diagnosed

No Natural History data

No clinical trials

No access to new treatments





Duchenne untreatable ?





Asymptomatic phase?



TNO study



The presence of developmental delay was evident at 2 to 3 months of age, with a higher proportion of young males with DMD failing to attain milestones of gross/fine motor activity, adaptive behaviour, personal/social behaviour, and communication

Between 12 and 36 months of age, differences in the attainment of developmental milestones concerning gross motor activity increased with age .

We also found differences in developmental milestones concerning fine motor activity, adaptive behaviour, personal/social behaviour, and communication between 12 and 48 months of age

In conclusion



True carefree period does not exist

True asymptomatic period does not exist

Early diagnosis adds to being able to give the best possible care

Good parenting needs attention



Early diagnosis allows for early adaptation and intervention





It is evident that newborn screening for Duchenne muscular dystrophy has been an area fraught with difficulties and challenges. However, with many families reporting that an early diagnosis allows better planning or call it better parenting – and with novel treatments now having received regulatory approval - now is the time to address these challenges to ensure newborn screening for the condition can be put in place.