Neonatal screening: Contribution of the French National Rare Diseases Data Bank to Estimate Early Mortality of Affected Patients

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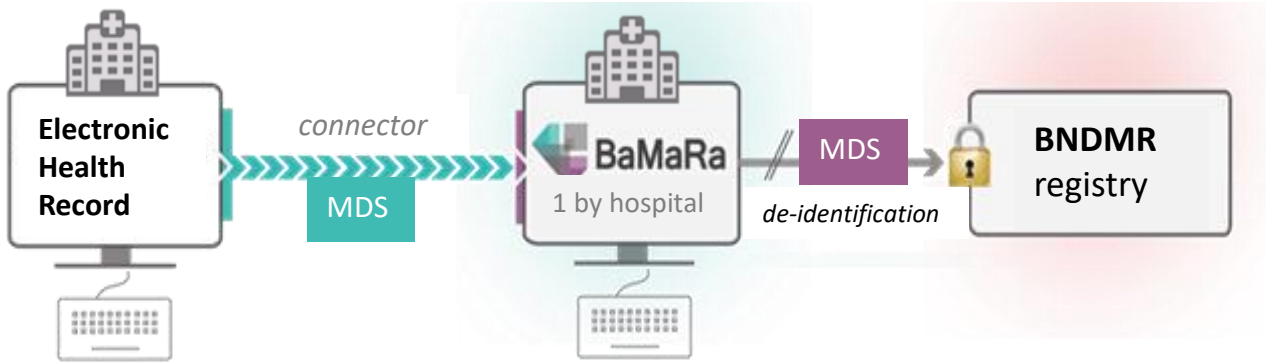
In 2019, France was lagging behind significantly with only 6 pathologies detected:

- Phenylketonuria
- Cystic fibrosis
- Congenital adrenal hyperplasia
- Congenital hypothyroidism
- MCAD deficiency
- Sickle cell anemia under certain conditions

7 new diseases in 2022:

- Isovaleric aciduria
- Type I glutaric aciduria
- LCHAD deficiency
- Carnitine transporter deficiency
- Homocystinuria
- Maple syrup urine disease
- Type I tyrosinemia

All pathologies with neonatal screening are rare diseases and therefore affected patients are recorded in the rare disease information system of the French National Rare Disease Data Bank



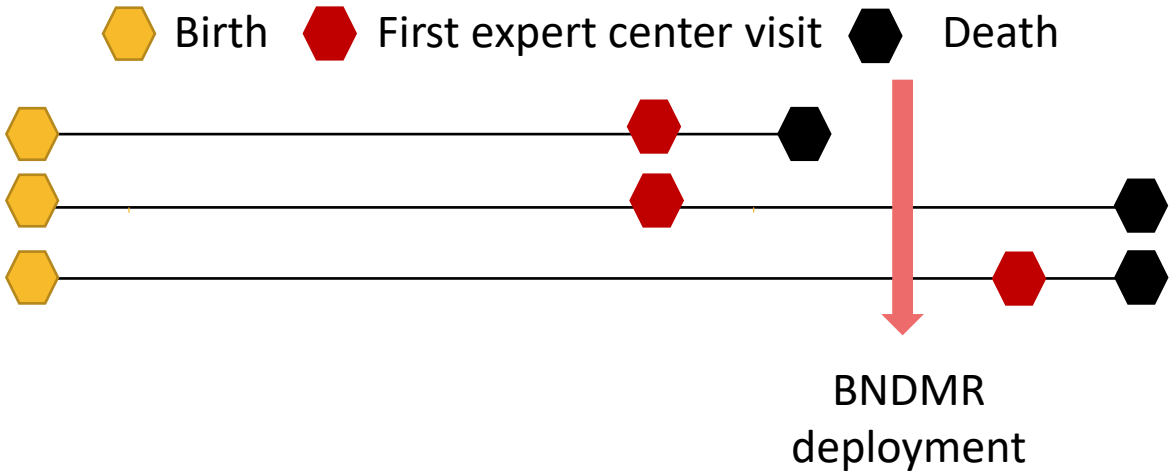
The RD French minimum data set (MDS)

The French MDS contains about 60 items, including :

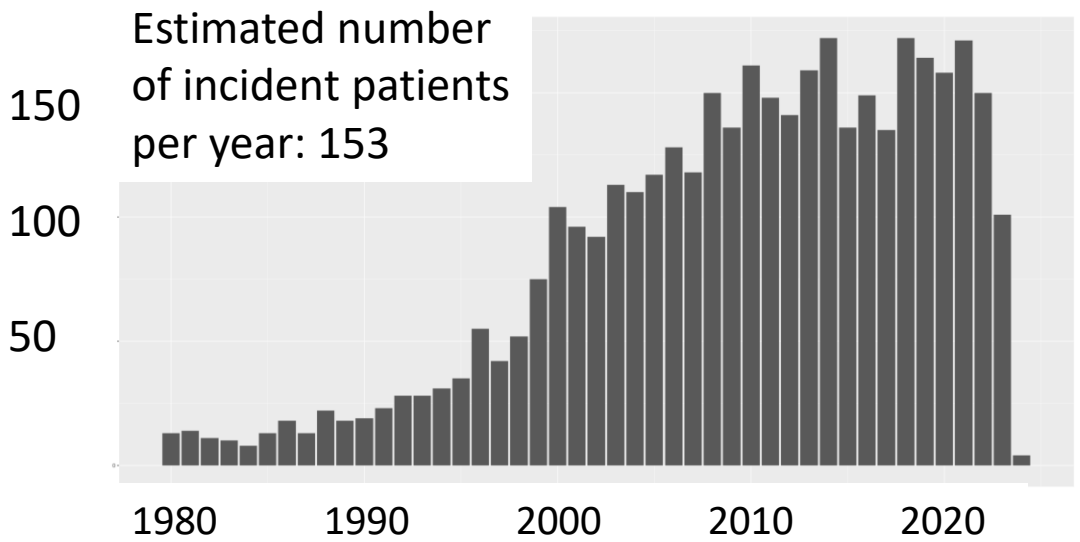
- Patient identification
- Vital status
- Care pathway & activities
- Ante and neonatal data
- Treatment (orphan drugs)
- Diagnosis (ORPHAcodes)
- Diagnostic history and accuracy
- Phenotype (HPO / ICD-10)
- Genotype (HGNC / HGVS)

Estimation of the number of incident case per year

- **How to estimate the birth incidence of different pathologies?** Normally, for genetic diseases, we should observe a nearly constant number of births per year. In fact, this is not the case for two reasons: we have only collected data since 2010, so many patients born in the 1980s and 1990s were already deceased, resulting in an underestimation for those years. For recent years, patients have not all yet shown the first signs of the disease or are in medical wandering, and therefore have not consulted an expert center yet.



Congenital hypothyroidism



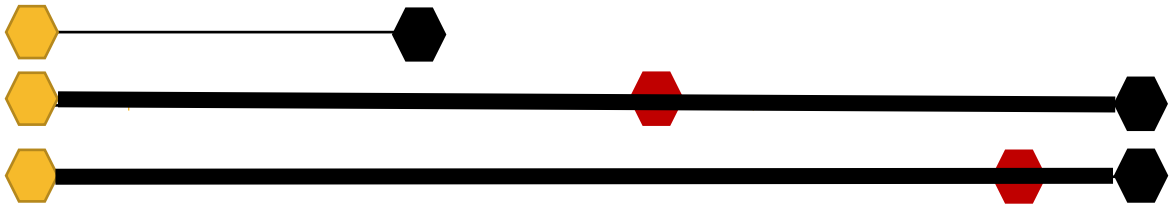
Number of estimated incident patients per year

- Phenylketonuria: 90
- Cystic fibrosis: 209
- Congenital adrenal hyperplasia: 75
- Congenital hypothyroidism: 153
- MCAD deficiency: 28
- Sickle cell anemia: 319
- Isovaleric aciduria: 5
- Type I glutaric aciduria: 7
- LCHAD deficiency: 8
- Carnitine transporter deficiency: 4
- Homocystinuria: 5
- Maple syrup urine disease: 9
- Type I tyrosinemia: 7

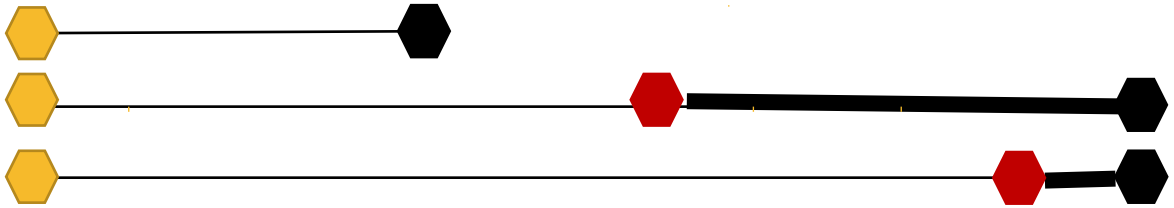
Survival estimation

Birth First expert center visit Death

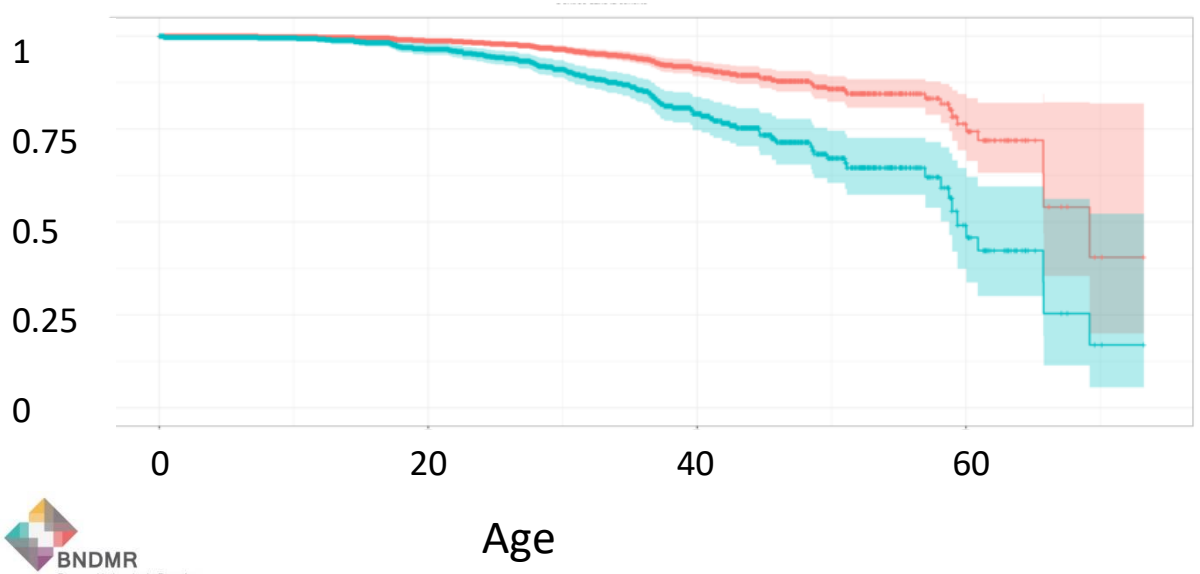
First option (blue): survival estimated by considering that each patient is followed up since birth (survivorship bias)



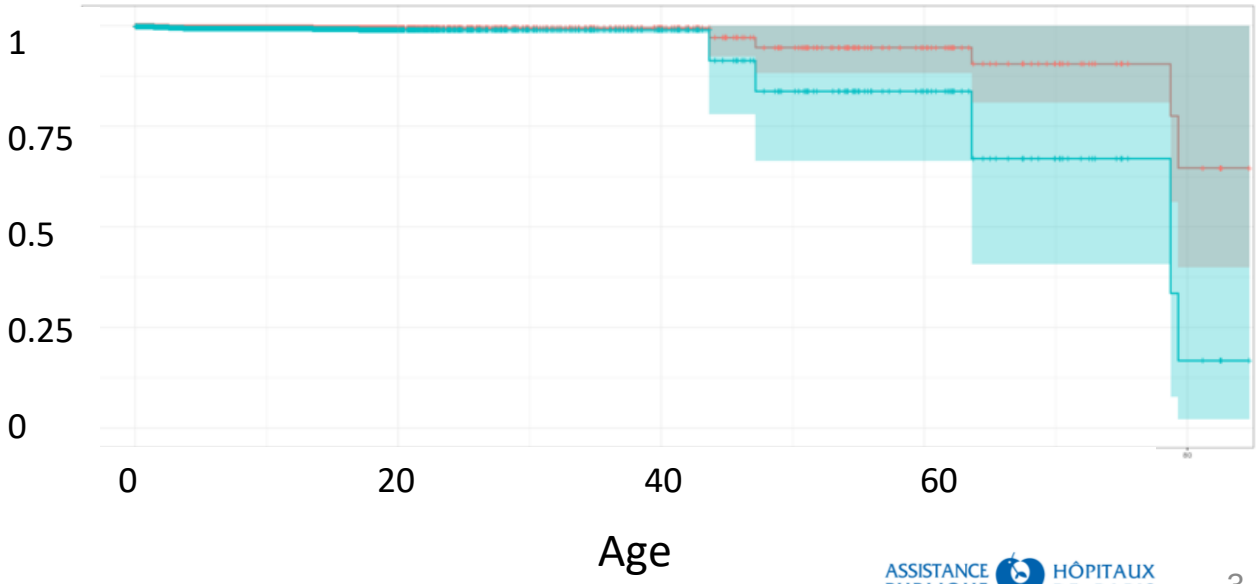
Second option (red): survival estimated by considering that each patient is followed up since first expert center visit (left truncation)



Cystic fibrosis survival estimation



Congenital hypothyroidism survival estimation



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