

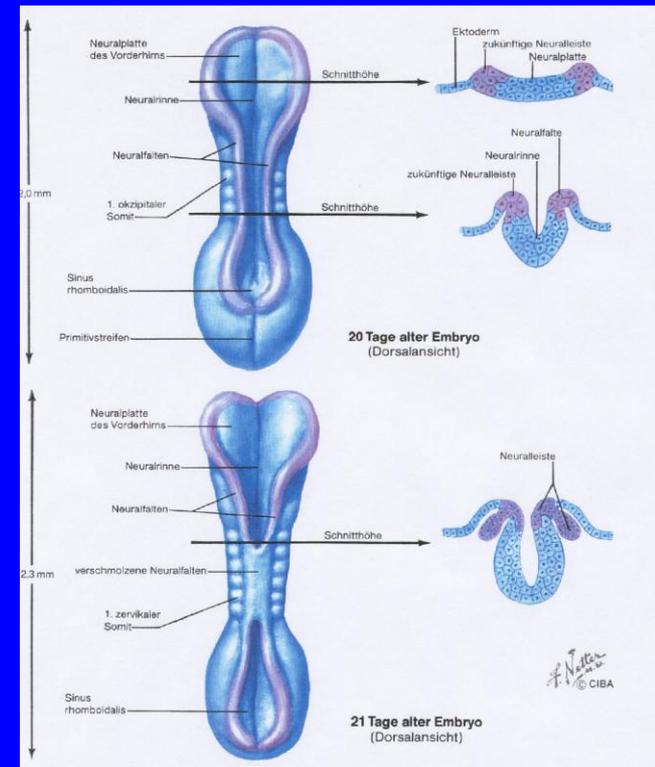
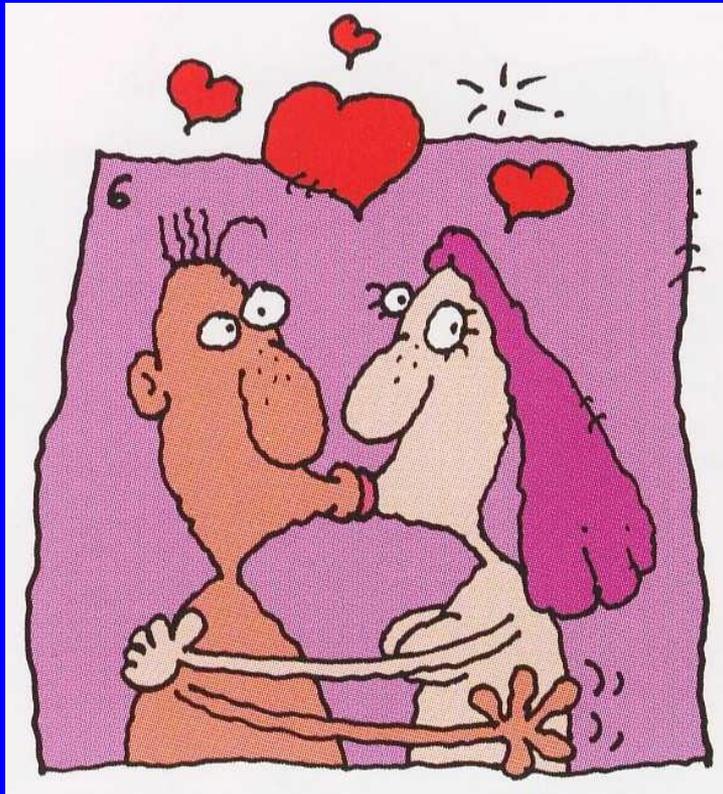
Successing care of children with Neural Tube Defect

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2013

Brussels, 29 June 2011 EUROCAT data
**Neural Tube Defect is in Europe most
common birthdefect**



The majority of neural tube defects (approximately 70%) occur in isolation and are said to show multifactorial inheritance

Up to 24% of neural tube defect cases occurs in association with other congenital (incl. cytogenetic) abnormalities



- It was the custom when a NTD child was born with opened spina bifida in a complex with a neurological deficit or hydrocephalus, he was considered incurable and the best way to resolve the problem was to send the child to an orphanage

- Spina bifida and hydrocephalus patients had been considered as non-treatable cases in Estonia for decades
- This situation changed in 1985, when we decided to start providing contemporary treatment to SpBH patients

When we started the idea was to deal only with neonates (it still is only an idea)

In Estonia paediatric specialists treat children up to the age of 18

- Childrens come to the children`s hospital straight from the maternity hospital
- The child is given complex care

The child is given complex care



We have to introduce handling these patients as teamwork in addition to our daily work



- Paediatric surgeon
- Urologist
- Orthopaedic surgeon
- Neurologist
- Psychologist
- Speech therapist
- Physiotherapist
- Social worker
- Orthotist

- At first we didn't include older children who had been declared incurable, but we soon found that it's possible to help these children and give them a better life - despite the late start



- However the contingent is small and opening a special outpatient clinic just for these patients would not be practical
- All Estonian Spina bifida and Hydrocephalus patients are under surveillance of our hospital

- When Estonia regained its independence , our politicians advised us to establish a non-profit organisation for the treatment of SpBH, as general understanding and opportunities were very limited behind the iron curtain



Estonian Spina Bifida &
Hydrocephalus Society

since 19 January 1991

www.kelluke.ee

IF SpBH president Pierre Mertens from Belgium visited Estonia in year 2009



- In the early years, this allowed us to obtain knowledge from our Finnish and Swedish colleagues
- By chance we managed to establish very close cooperation with the Charite Clinic in Berlin

- We are all very lucky, because we consider it to be one of the leading clinics in the world



Paediatric Surgeons take care also of brain tumors, craniosynosthosis and other rare diseases



We are thankful to our colleagues from neighbourhood and cooperation in different level

Incidence of NTD in Estonia is in decline

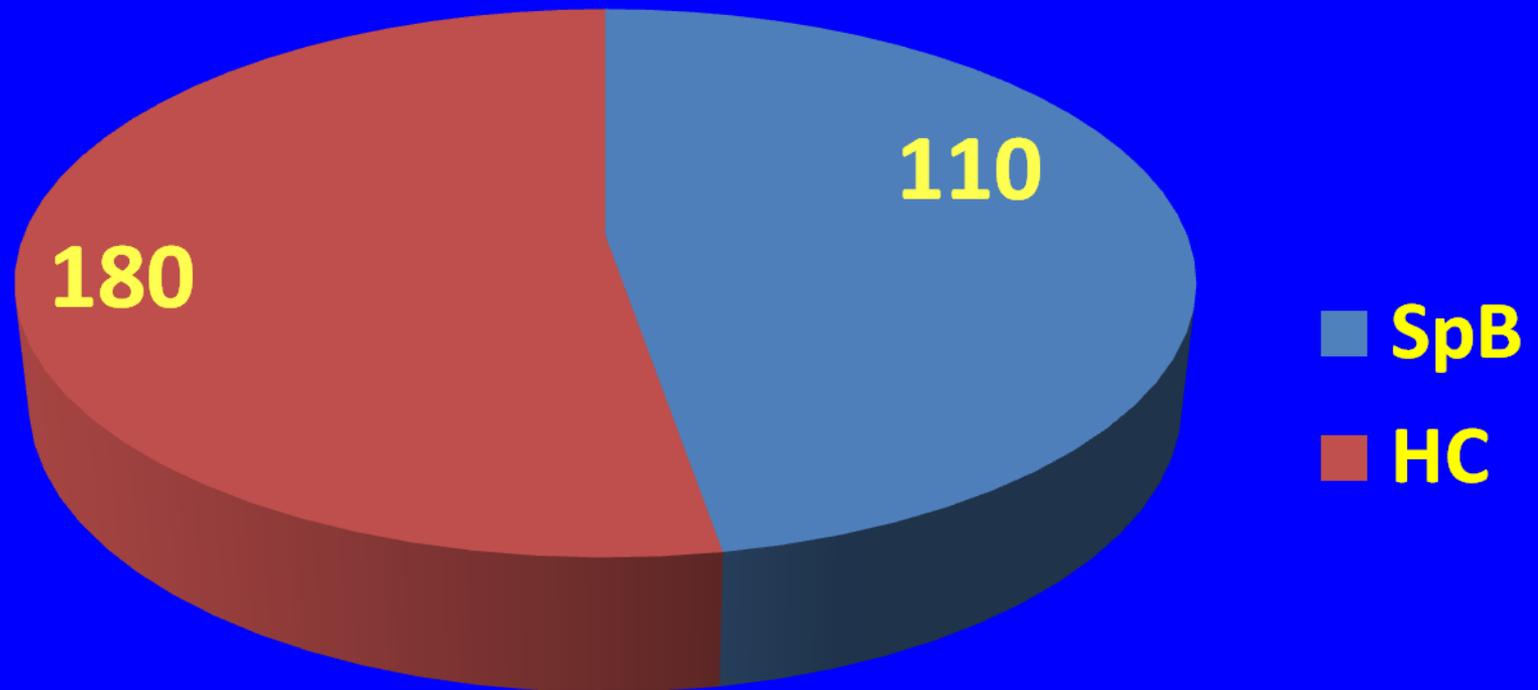
- In **1988** 22 SpB babies born in Estonia
there was birth rate the highest
ever 22 000
- In 2006 11 SpB babies born in Estonia
There was 11 647 abortion
and only ~8% from this were
spontaneous. Birth rate was 14 731
- In **2012** 0.7 per 1000 live-born babies
There was first year, when birth
rate was higher as abortion rate

- During the last 27 years we have collected 110 patients with isolated HC and 180 patients with spina bifida

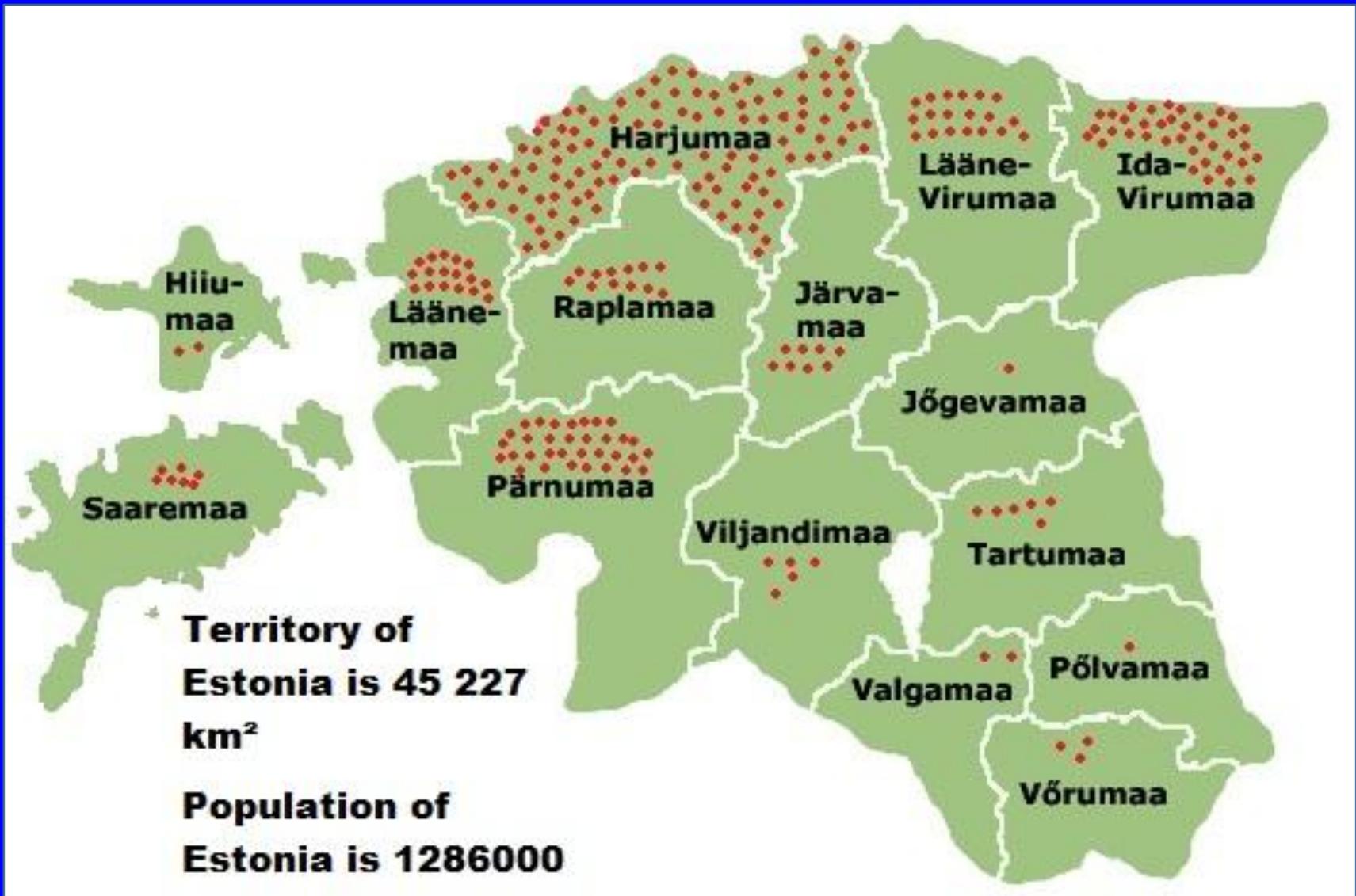


- Our main task is to establish a nationwide register of HC and SpBH patients consisting of medical records about treatment (interventions) and their outcomes in adult lifespan in order to improve their quality of life.

Estonian NTD Pathology Register as of 1 January 2012



SpBH regional spread



Decline in prevalence

Decline in prevalence



- **Socio-economic factors/awareness**
- **Prenatal care**
- **Folic acid fortification/
supplementation**
- **USG technique – induced terminations**

12/06/2012

Epidemiological studies have provided valuable insight for

- a) researchers to identify nongenetic and genetic factors contributing to etiology
- b) public health officials to design and implement policies to prevent NTD pregnancies and
- c) individuals to take precautions to reduce the chance of having an NTD-affected pregnancy

Cooperation with Estonian Genome Center

- When more heritable forms of human HC and SpB are identified and underlying genes and their functions are characterized, then this knowledge could be used to improve patient care in a variety of different ways such as prenatal diagnosis and new potential therapeutic approaches

- A new approach that captures the exons of all the genes in the human genome (whole exome) has been successfully used to identify causative genes for rare single-gene disorders

The first investigations show, that NTDs recur within families, with first degree relatives of an NTD patient possessing a 3–5% risk of having offspring with an NTD and second-degree relatives a 1–2% risk

- Over a hundred candidate genes have been examined for risk association to human spina bifida (SpB)
- Folic acid supplementation is an effective method for primary prevention of a proportion of NTDs in both humans and mice
- First steps with Folic Acid Prophylactics to prevent congenital abnormalities in Estonia have already been done



- Citizens of EU Member States will be able to travel to anyother Member State for treatment from autumn this year
- This will bring about new problems – we have to ensure that treatment principles and tactics are the same. We must not harm the patient, but give them as much treatment as they need. But how much do they need?

- It would be very welcome if EU Member States had common principles regarding NTD pathology
- There are already situations where the solution to an existing health problem depend on past treatment



Increase in survival

- **Evidence based guidelines**
- **Improvements in**
 - pre-, peri- & neonatal care
 - diagnostic procedures
 - neurosurgical techniques
 - urological care (CIC!)/urodynamics



12/06/2012

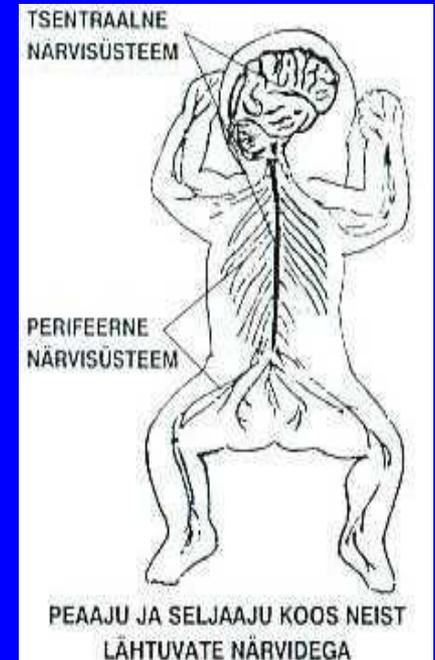
- Today we have ~60 young adults in various states of health
- The problems of our young adults are completely different from those of children, but we cannot arrange complete solutions for their problems yet

Example: Four girls have become mothers. It is good luck, that all had healthy babies. But all of mothers have had to solve serious problems

Problems of NTD Patients

- All Systems of the body,
but not for everybody

Slogan from the first SpBH Worldcongress
In USA, Orlando 2010

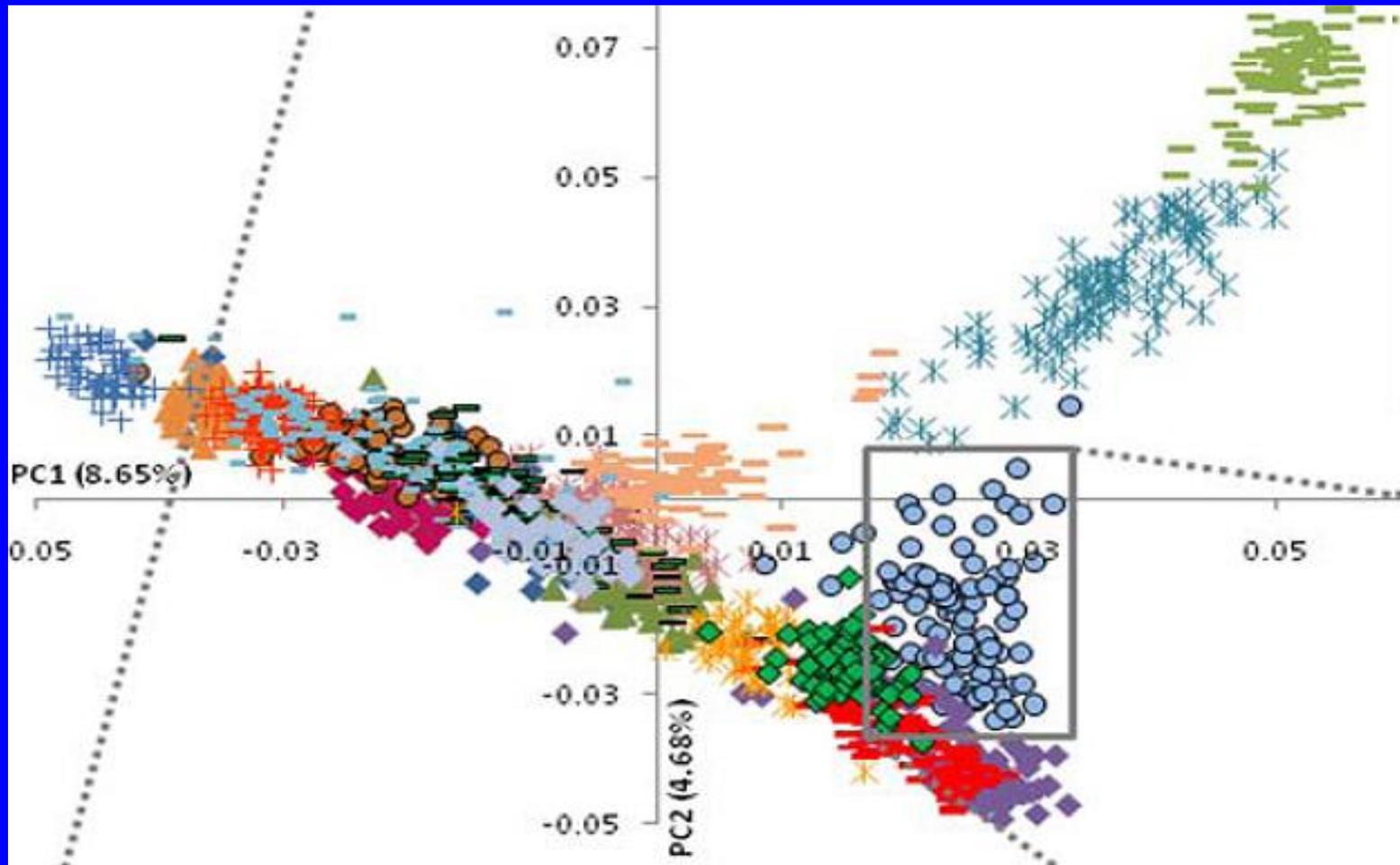


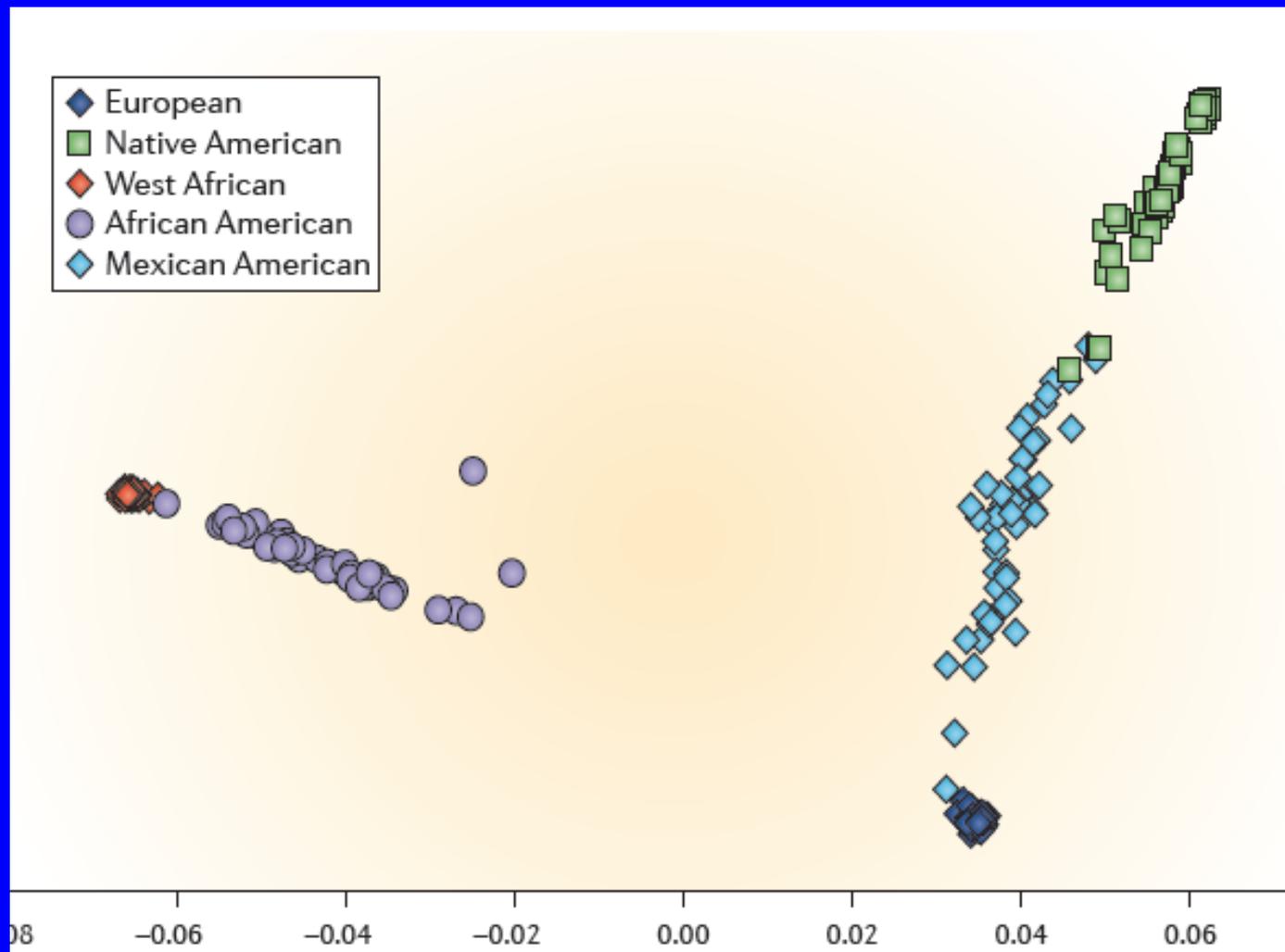
NOTHING
IN SPINA BIFIDA
GETS EASIER WITH AGE

Problems of young NTD adults

- Acquires helplessness is obvious in some of them, probably caused by us giving them excessive care or being too scared
- However should we even worry about the adults so much? Young people have their own plans – maybe we want to interfere too much with the lives of others?

European genetic map





Intercontinental populations: heritage

- Systemic approach involving clinical service development, genetic epidemiology research and training the professionals together with providing sufficient resources of prevention looks most promising to improve the healthcare and well-being of young adults affected with NTD phenotypes.

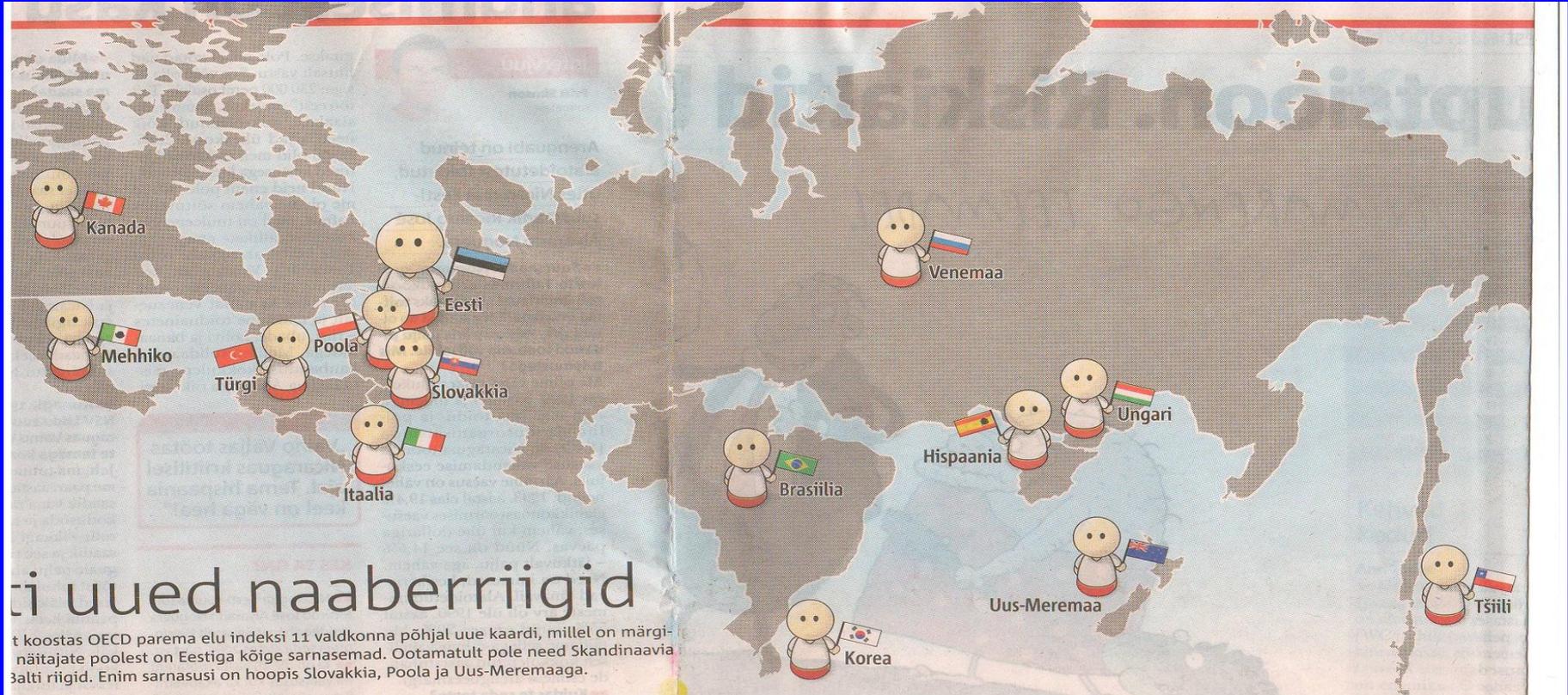
The Estonian Human Development Report was published in the media a month ago

- We prefer comparing ourselves to the wealthy Scandinavian countries rather than our close neighbours on the continent
- Estonia is like a lonely cross-country skier, who cannot catch those who are ahead and who is not threatened by those coming from behind

- The report indicated that we have a narrow view of the world that in many cases borders on Nordic countries and Latvia.



When we comes to quality of life, we are more similar to Estonia's new neighbours to Slovakia, Poland, Hungary, Chile, Brazil, South Korea



Eesti uued naaberrigid

...t koostas OECD parema elu indeksi 11 valdkonna põhjal uue kaardi, millel on märgitud näitajate poolest on Eestiga kõige sarnasemad. Ootamatult pole need Skandinaavia ja Balti riigid. Enim sarnasusi on hoopis Slovakkia, Poola ja Uus-Meremaaga.

Sissetulek ja jõukus	Töö- ja pere-elu tasakaal	Haridus	Keskonna kvaliteet
Mehhiko: 0,9	Uus-Meremaa: 6,4	Uus-Meremaa: 7,8	Hispaania: 6,6
Eesti: 1,3	Eesti: 6,5	Eesti: 7,9	Eesti: 6,8
Slovakkia: 1,3	Kanada: 6,5	Poola: 7,9	Itaalia: 6,8

Subjekttiivne heaolutunne	Töö ja palk	Tervise-seisund	Kogukonna-sidemed	Kodaniku-kaasatus
Kreeka: 1,5	Türgi: 2,6	Ungari: 3,9	Brasiilia: 7,4	Venemaa: 1,9
Eesti: 1,9	Eesti: 3,9	Eesti: 4,4	Eesti: 7,5	Eesti: 2,1
Poola: 2,5	Slovakkia: 4	Korea: 4,8	Itaalia: 7,7	Slovakkia: 3,1

Allikas: OECD Better Life Index (Eesti inimarenngu aruanne 2012/2013)

- One of the main messages of the report was that we cannot copy anyone else in terms of our lives and conditions, but we should rather develop our own model of quality of life
- Our situation is the worst in areas where we don't have a clear national policy, such as housing and healthcare
- The report highlights that it is time to start helping the weakest members of society

Perhaps we should use the opportunities we have in Estonia – for example gene research – to help us better understand ourselves and to improve our lifestyles, learn new knowledge and skills

Thank you for your attention