The Neuronal Ceroid Lipofuscinoses (NCL) in Flanders, Belgium

Starting from the available individual and demographic data, we aim to evolve towards a standardized multidisciplinary approach. This can facilitate early diagnosis and better support for all concerned

Katrien Keppens, MD[1], Geertrui Peirens, MD[2], Lieve Standaert, MD[1], Stefan Mattheeuws, MD,parent[3]

1 De Kade, Spermalie, Bruges, Belgium

2 Ganspoel, Huldenberg, Belgium

3 ContactpuntNCL, Mol, Belgium

www.contactpuntncl.be

katrien.keppens@de-kade.be

EPNS 2013

10th European Paediatric Neurology Society Congress 2013

25 - 28 september 2013

Square - Brussels Meeting Centre - Brussels, Belgium

Juvenile NCL: symptoms

Age in years

Gradual visual loss

Learning difficulties

Epilepsy

Changes in behaviour

Disorder in:
- motion
- sleep
- speech
- psyche

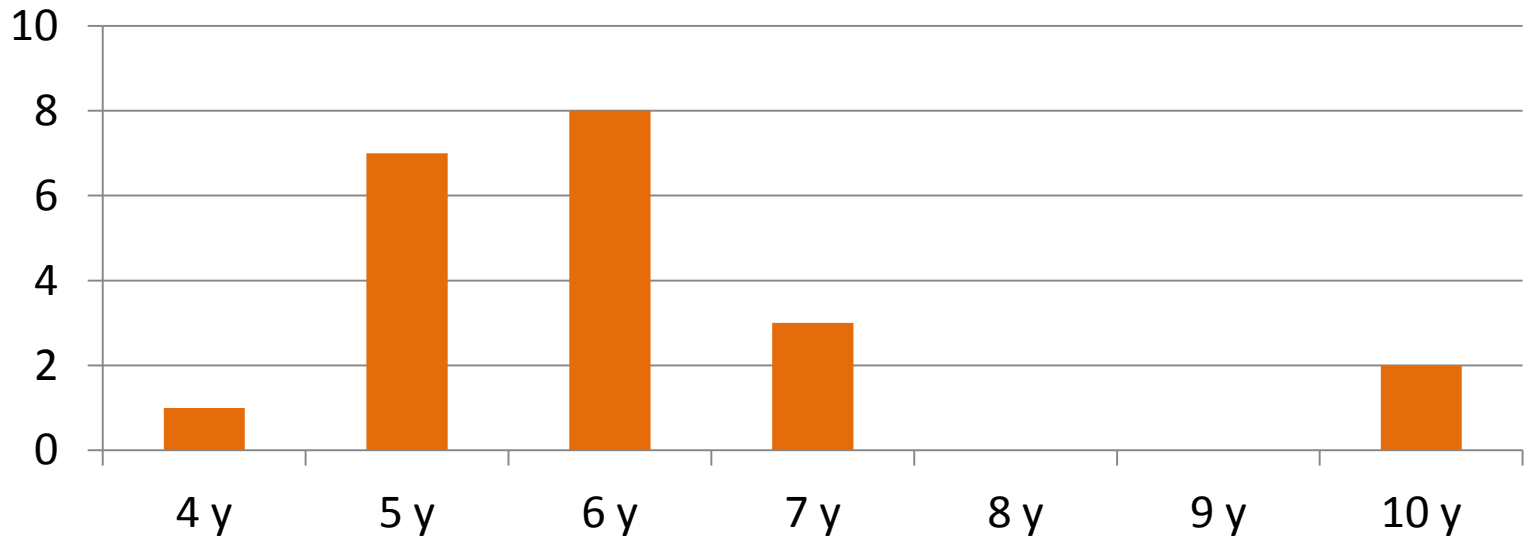
Wheelchair bound
More dependent
Refractory epilepsy
Difficult to understand

Aphasia
Vital function failure
Tube feeding
Bedridden
Palliation

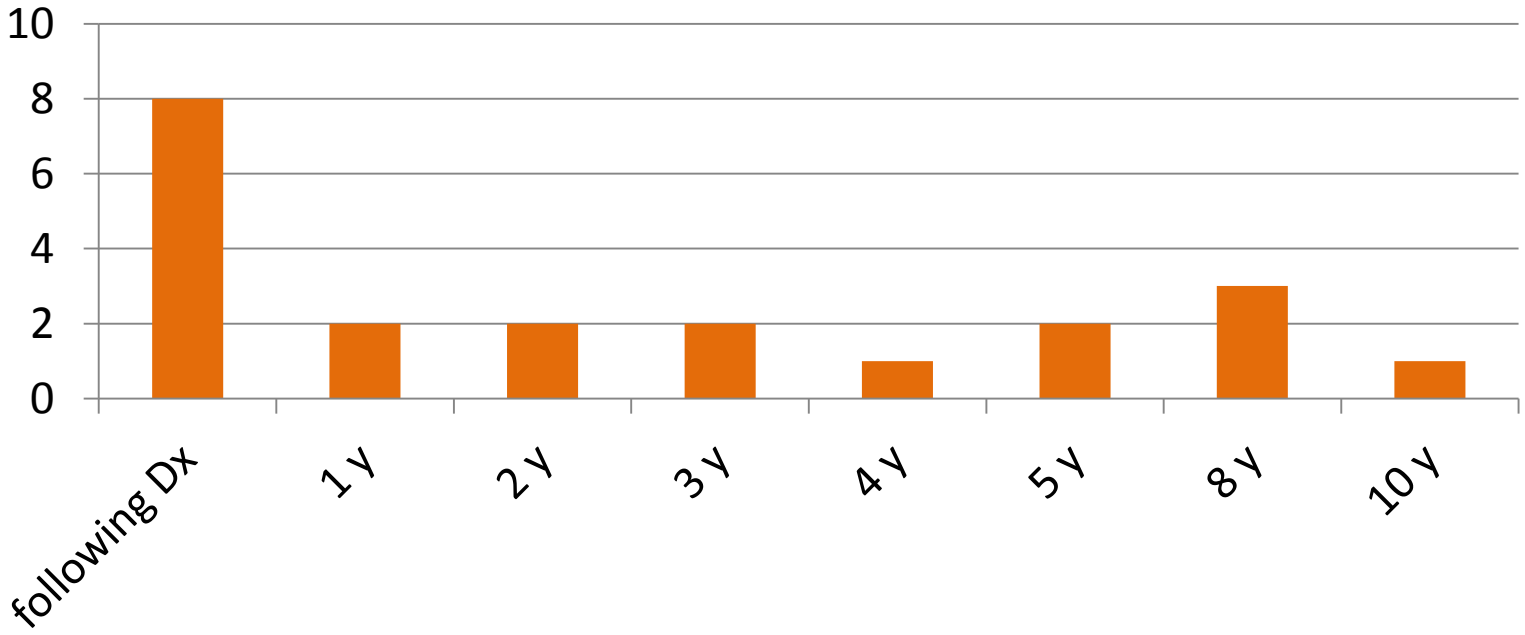
Retrospective results:
Available individual and demographic data

- 21 patients born between 1964 and 2004 with juvenile NCL
- Presenting symptom (21/21): gradual visual loss due to retinal dystrophy, ERG extinguished
- 3 families have 2 children with NCL
- 9/21 died:
  - 7 between 25 and 35 year of age due to NCL
  - 1 malignant neuroleptic syndrome (14 years),
  - 1 traumatic (11 years)
- Diagnosis:
  - before 1990: by demonstration of vacuoles in lymfocytes
  - after 1990: genetic testing (DNA sequencing)
- DNA , 10 patients:
  - 6 with 1 kb del (*CLN 3)*
  - 2 no DNA diagnosis (twins)
  - 2 unknown

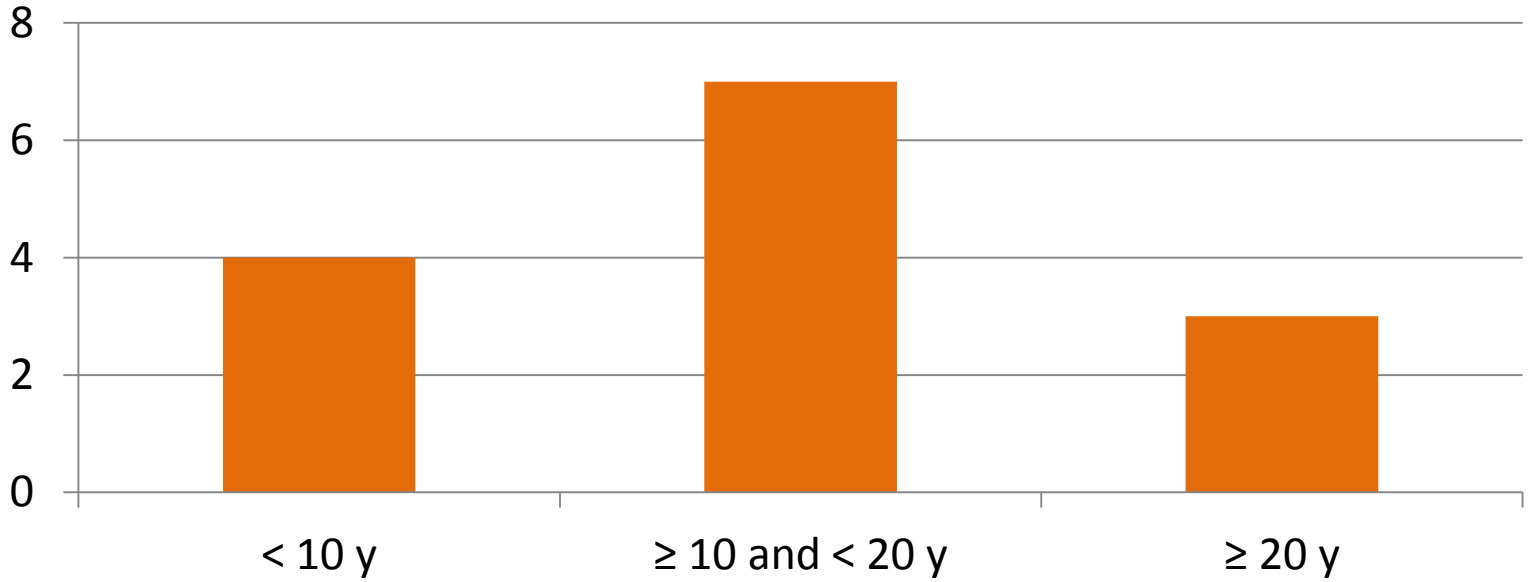
Presenting symptom = gradual visual loss due to retinal dystrophy around the age of 6 years



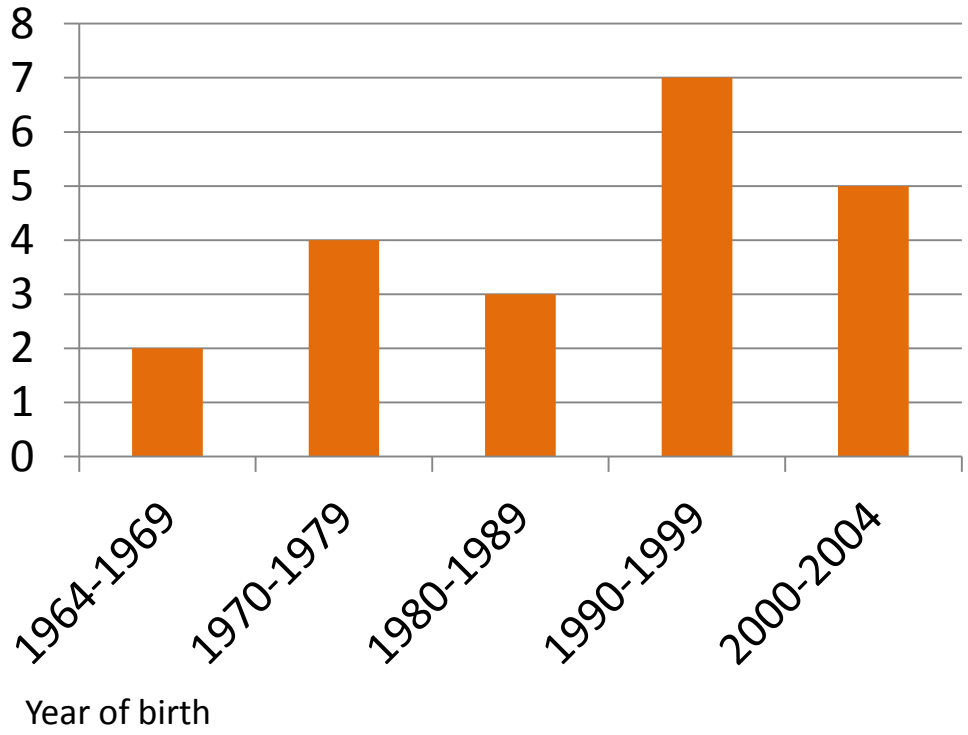
Time between presenting symptom and diagnosis



First epileptic insult



Estimated incidence of juvenile NCL in Flanders



21 patients born over a period of 40 years

Special education for blind = Ganspoel or De Kade/Spermalie

Life birth rate in Flanders (2010) : 70 079

Estimated incidence:

0,5/70 000 = 0,7/100 000

(United States: 0,7/100 000)

(Finland: 4,8; Norway: 3,7; Sweden: 2,2$_{/100\,000}$)

Objectives for the future

Establishing a standardized registry as a first step

- To increase the awareness for NCL
- To include all patients with NCL (not only juvenile form) in the registry
- To facilitate exchange of experience
- Towards earlier, accurate and complete diagnosis
- Towards a better multidisciplinary approach
  - Including professional caretakers
  - Including Contactpunt NCL (patient organization)
- To offer better support to patients and next of kin

Axial diagnostic classification for the registry

- Axis 1: affected gene
- Axis 2: mutation diagnosis
- Axis 3: biochemical phenotype
- Axis 4: clinical phenotype
- Axis 5: ultrastructural features
- Axis 6: functionality
- Axis 7: other remarks

Mole et al, The Neuronal Ceroid Lipofuscinoses (Batten Disease) 2nd Edition; Oxford UP, 2011

Conclusions

- A history of gradual visual loss in an otherwise healthy young child (4-7y) is very suggestive of juvenile NCL
- Through the registration we will gather all types of NCL
- A multidisciplinary approach guarantees better quality of care
- Medical doctors, paramedics, educators as well as parents, should be able to participate in this multidisciplinary setting