Keynote 1 Jan Veldink

Introduction

- Jan Veldink
  - Lead researcher project Mine
  - UMC Utrecht — Neurologist
- Topic — What is ALS – Are we on a verge of a breakthrough?
- How to solve ALS
  - 150 years of research
  - Challenging to say this will happen
  - But 'impossible' is not in my dictionary

What is ALS?

- Life time risk 1 in 300 people
- Over 400,000 patients worldwide
- Progressive muscle weakness
- Otherwise healthy/"fit" people
- Lethal disease (3-5 years)
- Most frequent reason physician-assisted death
- Video What is ALS
  - 3-5 years death
  - No cure for ALS
  - Course of disease
    - Change in DNA
    - Defective proteins
  - Protein aggregation
    - Defections
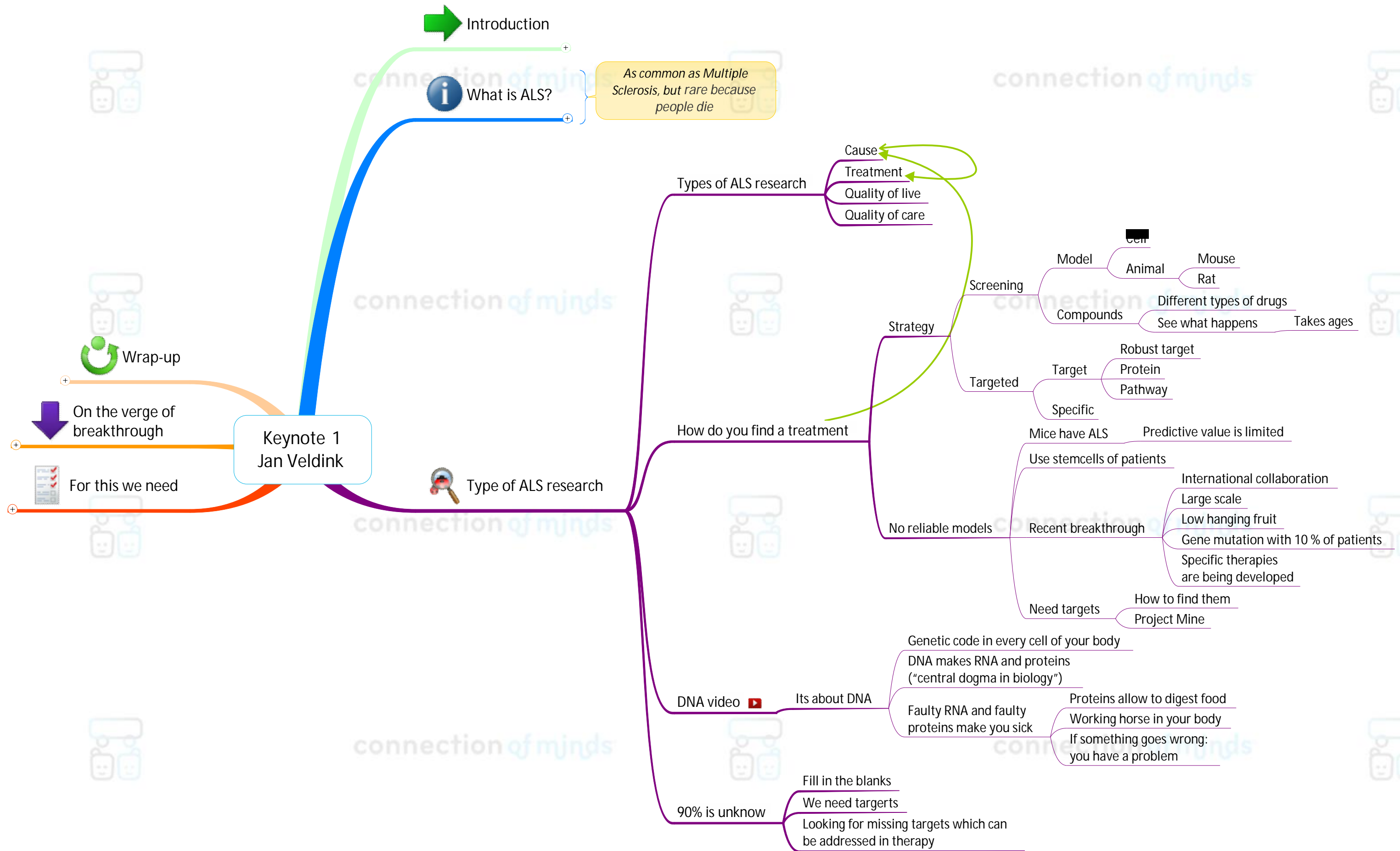   - Loss of muscle innovation

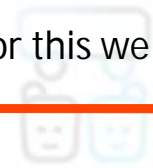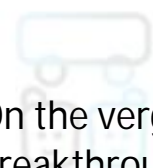*As common as Multiple Sclerosis, but rare because people die*

Type of ALS research

Wrap-up

On the verge of breakthrough

For this we need

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Types of ALS research
- Cause
- Treatment
- Quality of live
- Quality of care

Type of ALS research

How do you find a treatment

Strategy
- Screening
  - Model
    - Cell
    - Animal
      - Mouse
      - Rat
  - Compounds
    - Different types of drugs
    - See what happens
      - Takes ages
- Targeted
  - Target
    - Robust target
    - Protein
    - Pathway
  - Specific

No reliable models
- Mice have ALS
  - Predictive value is limited
- Use stemcells of patients
- Recent breakthrough
  - International collaboration
  - Large scale
  - Low hanging fruit
  - Gene mutation with 10 % of patients
  - Specific therapies are being developed
- Need targets
  - How to find them
  - Project Mine

DNA video ▶ Its about DNA
- Genetic code in every cell of your body
- DNA makes RNA and proteins ("central dogma in biology")
- Faulty RNA and faulty proteins make you sick
  - Proteins allow to digest food
  - Working horse in your body
  - If something goes wrong: you have a problem

90% is unknow
- Fill in the blanks
- We need targerts
- Looking for missing targets which can be addressed in therapy

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Wrap-up
- Unique
- Different time
- Lots of help from Accenture

On the verge of breakthrough
- Large scale research
- Pricing model
  - Number of DNA's
  - $$ per genome will be lower
- Project mine
  - Website
  - Donation

For this we need
- Strong international collaboration
  - Think global
  - Think big
  - Need complete picture
- ✔ Detailed characterized patients
- ✔ DNA samples
  - Good quality
- ✔ Healthy controls
- ✔ Technology
  - Store info
- ✔ Translational potential
- ✔ Bioinformatics expertise
- Funds..

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