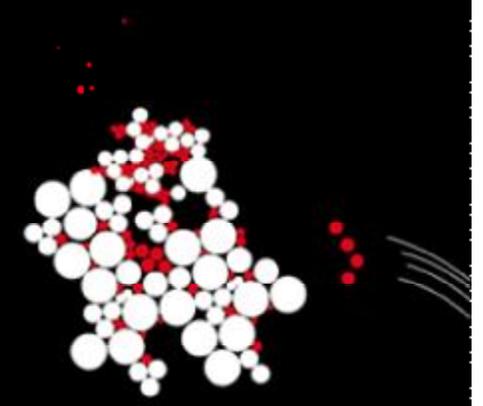


UNIVERSITY OF TWENTE.

Society & GENOMICS
Centre for Society and Genomics



Emerging stabilization in genomics.
Governance and practices of valorization

Tentative Governance of Emerging
Science and Technology, Enschede,
October 28-29, 2010

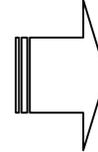
Roel Nahuis
Dirk Stemerding
University of Twente
Science, Technology and Policy Studies
r.nahuis@utwente.nl



The emergence of genomics in the field of clinical genetics

Clinical genetics

- § **Focus on monogenetic diseases**
- § **national networks of scientists, clinicians, funding bodies based around hospitals**
- § **clinical genetics centres linking laboratory research and diagnosis with clinical patient care and counselling**
- § **strong orientation of research to clinically relevant genetic diseases**
- § **important role for patient groups as intermediaries**



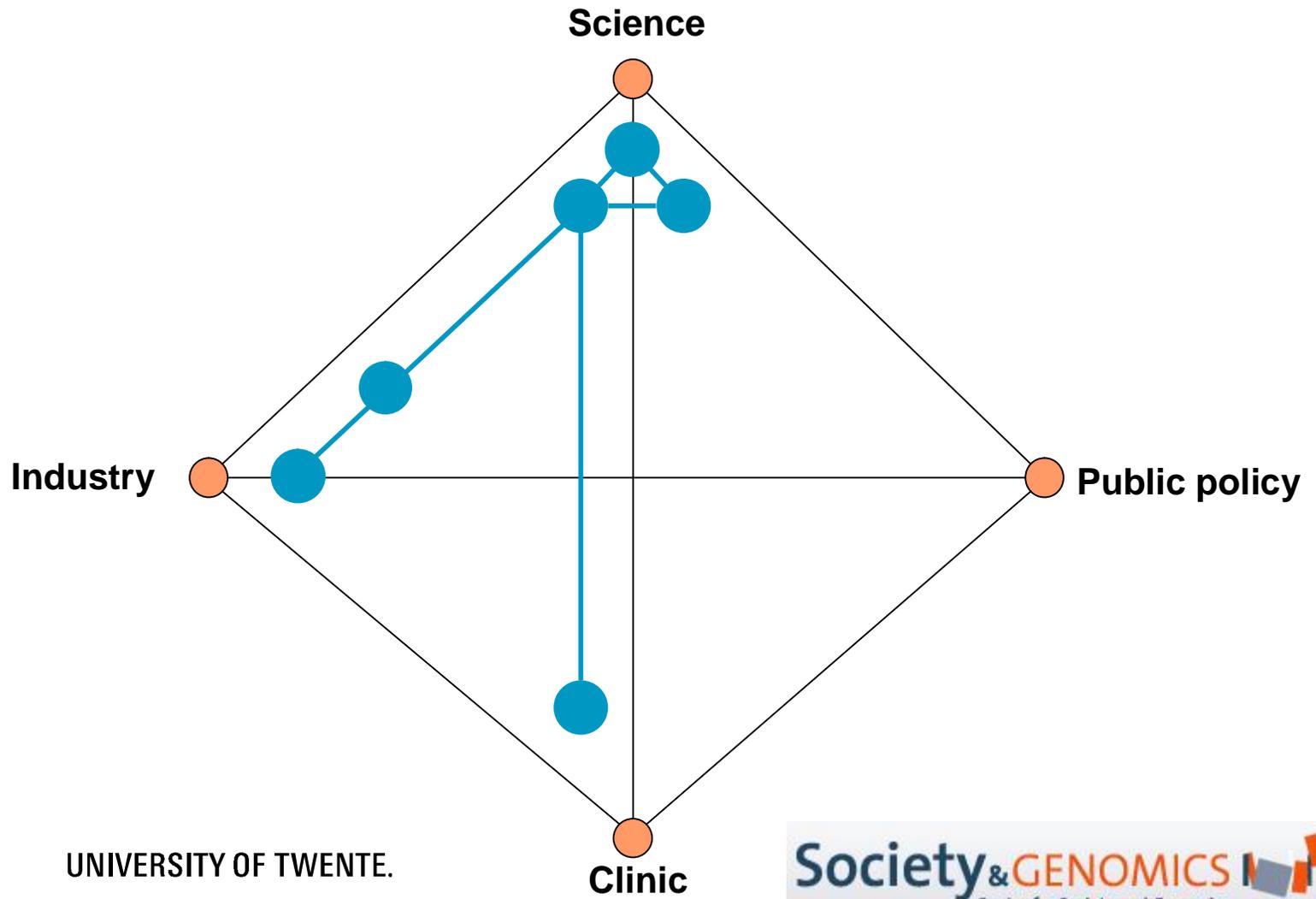
Genomics

- § **Focus on multifactorial diseases**
- § **large-scale consortia with international, multi-disciplinary collaboration**
- § **strategic public investments and public-private relationships**
- § **use of high throughput technologies**
- § **genetic databases as platforms linking academic and commercial interests**

Genomics as a new innovation regime

- § Emergence of genomics involves an epistemological and institutional transformation of knowledge production in human genetics
- § This transformation also has implications for the ways in which science and society are related in genomics
- § ‘Valorisation’ as a new challenge in genomics
 - § *“to ensure that society and economy benefit from the breakthrough enabled by genomics in important fields like health, sustainability, enabling technologies and society” (NGI homepage)*
 - § Measured in quantities of dissertations, patents, start-up companies, industrial matching
- § Narrow economic definition leads to discrimination of particular research fields (eg. does not appreciate contribution to new diagnostic criteria)

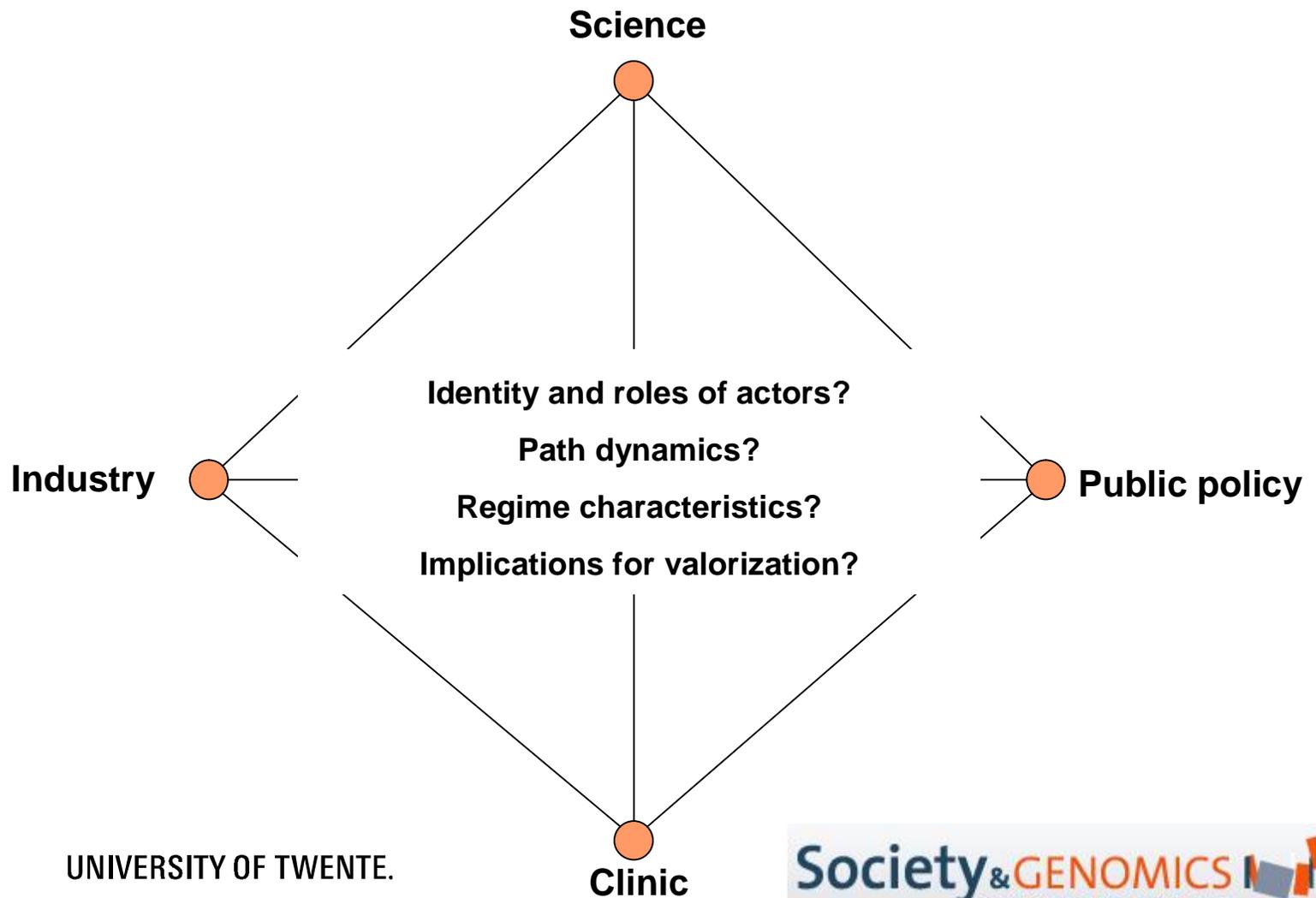
The multi actor world of innovation



Some useful concepts

- § Innovation regime refers to rules, procedures, and heuristics that guide activities and interactions in networks of research and development
 - § Resource driven regimes characterized by the exploration of multiple possible paths
 - § Technological regimes characterized by actor's beliefs about 'not yet exploited opportunities' along a specific path
- § Socio-technical paths refers to the (emerging) alignment between activities of actors at different poles (science, industry, clinic)
- à Valorization as knowledge production and uptake along paths towards clinical applications

The multi actor world of innovation



Two case studies

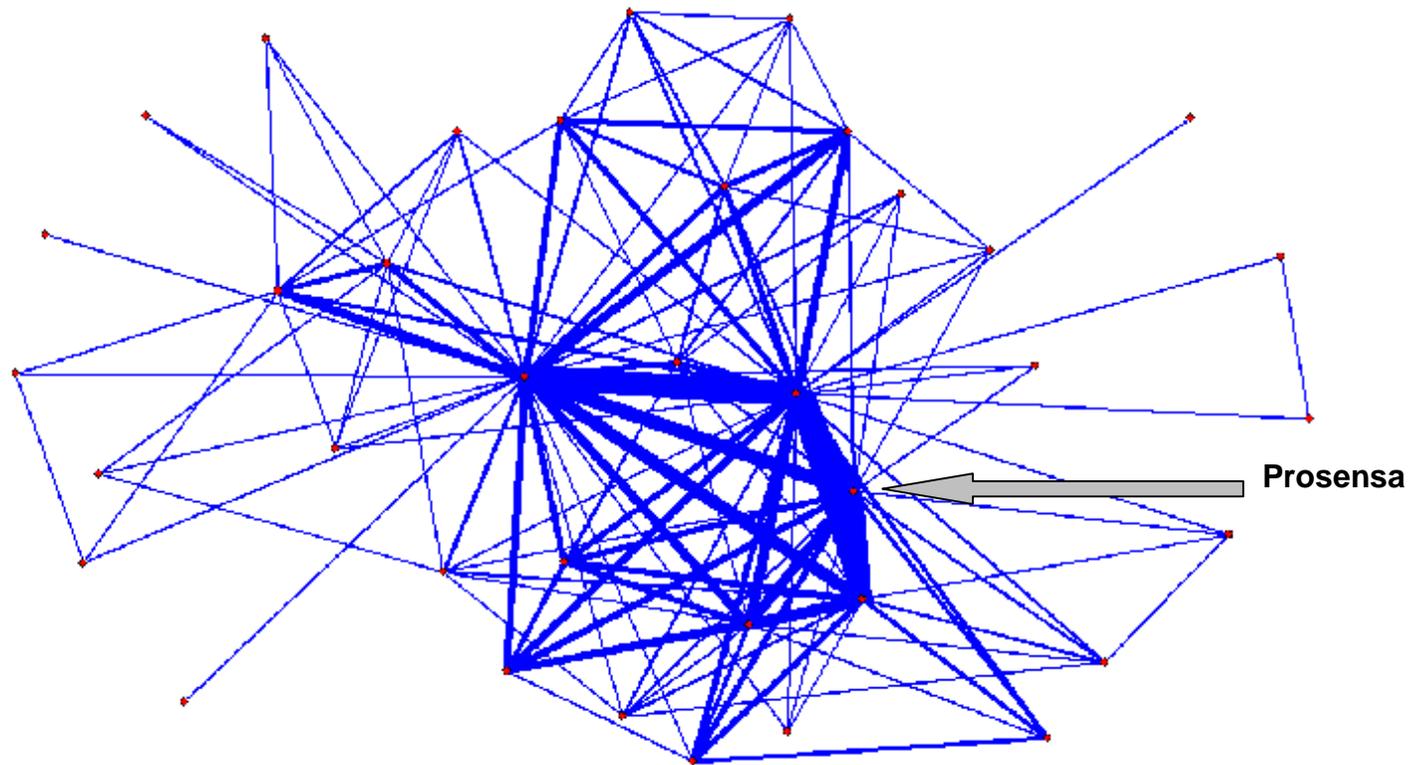
§ **Duchenne Muscular Dystrophy**

- § Major object of research in the clinical genetics regime (Nelis, 1998): monogenetic disease, strong patient involvement
- § Illustrates evolution of clinical genetics regime (e.g. current focus on therapeutic interventions w/ commercial actors)

§ **Alzheimer's disease**

- § Typical genomics regime: multifactorial disease, use of large biobanks, association studies as emerging paradigm
- § Emerging regime: merging of different activities (epidemiological and genetic research) and new institutional forms (genetic epidemiology departments)

Co-author map of Duchenne Muscular Dystrophy research



Topic = Duchenne
ISI classification = genetics & heredity
Address = Netherlands
Time: 2000-2010
Top 5 authors
>2 co-author relations

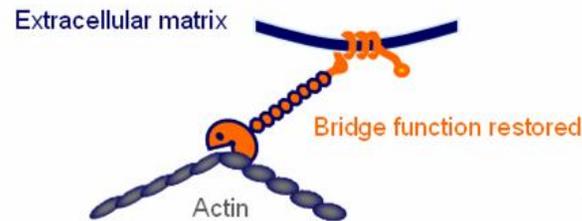
Exon skipping

Duchenne: Open reading frame disrupted
Truncated, non-functional dystrophin



small synthetic antisense oligoribonucleotides (AONs)

AON treatment: Exon skipped from pre-mRNA
Reading frame restored
Internally deleted, partly functional dystrophin
Becker-like phenotype



<http://www.humgen.nl/lab-aartsma-rus/>

Preliminary characterization of DMD regime

- § Actors: Center for Human and Clinical Genetics (LUMC), Prosensa, GlaxoSmithKline, DMD patients/parents/organisations
- § Path dynamics: alignment of scientific and industrial actors via patent, spin-off Prosensa, exon skipping technology
- § Innovation regime driven by the prospect of a new therapy
- § Implications for valorization:
 - § Science-industry relations: scientists' commercial ties, intellectual property, societal benefits of public funding, distribution of risks?
 - § (Clinical implications: exon skipping implies patient stratification → solidarity in patient organisation, orphanisation vs. clinical trials?)

Preliminary characterization of Alzheimer regime

- § Actors: epidemiologists, geneticists
- § Path dynamics
 - § Explorative, multiple possible paths
- § Resource driven regime: availability of abundant data and genotyping technology, complex agenda, hypothesis generation and testing
- § Implications for valorization:
 - § If one would equally appreciate the work done in fields like Alzheimer's genomics, then another notion of valorization is required: one based on the possibility of 'path creation' instead of 'path dependency'
 - § Management of expectations, co-creation of scenarios, interactions between different paths (diagnosis, detection, prevention, therapy), knowledge uptake, long-term resource availability

To wrap up

- § Emergence of medical genomics in the field of clinical genetics
- § Clinical genetics and medical genomics are governed by different types of regimes, with implications for valorisation policy
- § Economic valorisation feasible in case of established technological regime (clinical genetics regime of Duchenne research)
- § Beyond narrow definition: Valorisation as mid- and long-term accountability of path creation
 - § Legitimacy of possible scenarios of future developments