Abstracts for Innoboot 2020; Precision health care (October, 27 Online)

Jeske Timmermans **Sophie Dusoswa** **Roche**	**Precision oncology, the only way is up**
With increasing world population, increasing age, and increasing prevalence of chronic disease the current course of healthcare is unsustainable. The convergence of revolutions in data science and “omics” in life sciences provide an exceptional opportunity to transform healthcare. By increasing use of technology, like DNA sequencing, and also advanced analytics we aim to deliver data-driven, insights generating and value-based healthcare solutions designed to meet the needs of the individual patient, at a faster pace and at a lower overall cost to societies.	

Sharon Kolk **Tjitske Kleefstra** **Donders Instituut / RU / RUMC**	**Rare genetic syndromes and the path to personalized health care**
Neurodevelopmental disorders (NDDs) comprise a heterogeneous group of disorders with a large social impact. Frequently, similar genes appear affected by rare variants across NDDs and psychiatric conditions. The steep increase in gene discovery in these so-called Mendelian syndromes, syndromes caused by rare monogenic variants, allows us to study underlying neurodevelopmental biology and detailed clinical phenotyping. The implementation of tailored intervention for rare ND syndromes is the highly mandatory next step that should be taken after genetic diagnosis. We thereby investigate the consequences of a known genetic deviation on neurobiology, neurocognition and behavior (*bottom-up*) rather than investigate heterogeneous groups on certain neurobiological aspects (*top-down*). Our multidisciplinary Radboudumc expert center for rare genetic NDDs is working hand-in-hand with various research groups on campus and in the Netherlands as well as with various societal stakeholders to continue studies from syndrome discovery to development of etiology-based strategies for personalised management/therapy. One example of such an interdisciplinary consortium is the NWA project *ProMiSe* in which we aim to understand the underlying neurocognitive and biological mechanisms which will open doors to investigate the possibility of therapeutic (early/preventive) interventions and subsequent improvement of care. Our first and foremost objective together with societal organizations is to accommodate the need in creating a Knowledge Platform that will include all information on the syndromes we study, clinical as well as scientific, in easy-to-understand terminology. We furthermore aim to enhance the development and implementation of novel intervention strategies for Mendelian disorders by obtaining fundamental insights in both the clinical and biological consequences of mutated genes that cause Mendelian syndromes. By achieving this, we also aim to provide the field with a template how to integrate fundamental insights in biological mechanisms, with cognitive and psychiatric profiling in distinct syndromes. In this way tailored intervention strategies targeted at the severe behavior and psychiatric problems that are frequently encountered can be developed.	

Wilma Göttgens **Radboudumc**	**x**
x	