Valorisation
Addendum Valorisation

I met Philip* in a facility for people with intellectually disabilities. At that moment, he was a man in his late forties and he functioned at a severely disabled level. I learned that he formerly had lived for years in a psychiatric indoor clinic because of a treatment-resistant psychosis. Also, it became clear to me that during his childhood, he suffered from a cleft palate (which was corrected surgically). When studying his medical records, I saw that during adolescence, he functioned at an intellectual level (IQ) of 80, and when I assessed him, he nearly reached an IQ of 20. Because of several symptoms, we thought it would be wise to do a genetic check-up, and he turned out to have a deletion on 22q11.2. The intellectual deterioration that he suffered was dramatic and, in the literature, never described. When discussing this with colleagues, who also worked with patients with an ID, I learned that they recognized this phenomenon. This was the beginning of the research leading up to this thesis.

We decided to focus on the 22q11.2 deletion syndrome (22q11DS) patients with a moderate-to-severe intellectual disability (IQ below 55). In the 22q11DS literature, it was said that these patients were rare. Within a year, we included in the Netherlands 33 patients with an IQ below 55. During this time, we came in contact with parents and family members of these 22q11DS patients, and many of them told us that nobody could tell them about what to expect in adulthood, how to treat and cope with the problems they suffered.

22q11DS is the most common microdeletion syndrome and occurs in about 1:2000 people (Shprintzen, 2008) and, recently, Grati et al. (2015) reported an incidence of 1:992 in a cohort of over 9500 pregnancies. Based on the first figure (1:2000) it is seen as a rare disorder (a disorder affecting fewer than 1 in 2000 (http://www.eurordis.org/about-rare-diseases), but the findings of Grati et al. (2015) suggest that 1:2000 is an underestimation and it no longer should be seen as a rare disorder. There has been a lot of research about several topics in this syndrome, and the majority of it is done in children, adolescents and young adults. Our knowledge is focused on these ages, but how symptoms evolve in adulthood is less known. The phenomenon deterioration in 22q11DS is neglected in literature and therefore not known by laypeople, and even by many professionals. The findings of the research described in this thesis will be very valuable for parents and professionals, especially concerning what to expect during life (prognosis).

Based on the number of births in the Netherlands in 2012 (173.000 newborns in 2012, source: http://www.nationaalompas.nl/bevolking/geboorte/huidig), there would be an annual rate of about 80–90 newborns with this syndrome. Based on the Dutch inhabitants (16,8 miljon), and the 1:2000 assumption, there would live
over 8000 patients with the 22q11 deletion syndrome in the Netherlands. With the latest findings of Grati et al. (2015), these numbers could be doubled. Of all 22q11DS patients, about one third will suffer from psychosis, especially schizophrenia. Those who suffer from chronic psychotic symptoms (most of the time, this will be diagnosed with the term “schizophrenia”) are the ones who are (probably very) vulnerable to a deterioration process. This is accompanied by personal suffering and often with a lot of desperation in the family, but also for professionals. Naturally, this also expresses the costs needed for the care of such patients.

Knowledge of the phenomenon deterioration and its psychopathological mechanisms can give us clues for possible treatment options. If new strategies are found, they can prevent a lot of suffering in patients and their families. Another aspect is that aforementioned knowledge can give clues for the treatment of other psychiatric disorders. If strategies (pharmaceutical or other strategies) are helpful in 22q11DS patients, it is in our opinion useful to investigate these strategies in other psychiatric disorders; for instance, schizophrenia or anxiety and mood disorders (both occur commonly in 22q11DS).

The reported research in this thesis are first steps towards a better understanding of people alive today with 22q11DS and designing appropriate healthcare for people suffering from this condition.

*This case is anonymized

References
