'What are you proud of?' 'Everything'

by Claudia Cannavo[*]

LonDownS Consortium is an association of researchers working on different aspects of Down syndrome. We celebrated World Down Syndrome Day 2018 by organising a day of activities, scientific talks, and games to thank people with Down syndrome that took part in our studies.

- The 21st of March (21/3) is World Down Syndrome Day – as people with Down syndrome have 3 copies of chromosome 21.
- The LonDownS Consortium is a group of researchers working on different aspects of Down syndrome, that come together to build a comprehensive approach on Down syndrome research.
- To celebrate World Down Syndrome Day 2018, LonDownS Consortium organised a day of talks, games, and activities for people with Down syndrome and their family and friends, aimed at bringing science closer to the non-scientific community

Research is often accused to keep itself separated from the ‘real world’. It is easy to imagine the stereotype scientist, white labcoat and pipette in his hand, mixing coloured potions that seem to be totally unrelated to the experience that people have in their everyday life. This is partially true; by focussing on the small problems to get to the big ones, it is easy for us scientists to lose the big picture. On World Down Syndrome Day 2018 we tried to bridge science and everyday life; as a way to say ***thank you*** to the people that allow research to move forward by taking part in our studies.

We organised a series of activities spanning from Neuroscience-related activities to talks to games. People with Down syndrome and their families, friends, and carers learnt about the different parts of the brain by wearing brain-shaped caps; built memory books; and got to know more about chromosome 21 using puzzles and games. Students just starting out on their research journey shared their passion for the brain with participants by using Play-Doh to model the way brain cells connect to think and create memories. Visitors could also attend talks in which scientists presented the results of their research. *How is it done? How is it useful? How will it help me?* The room was packed: everybody wanted to have a chance to see the scientists’ research progress, and to ask questions that Google could not answer.

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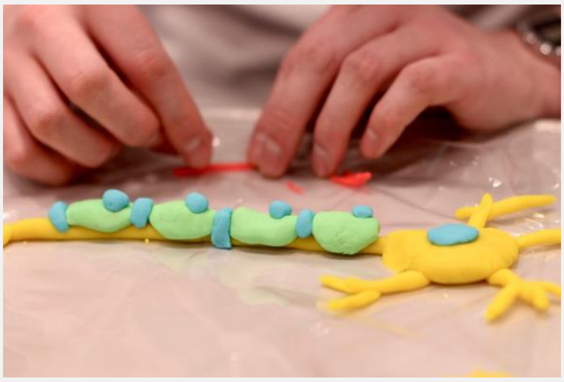

Hana, a researcher working with infants and toddlers with Down syndrome, spoke about the research at Birkbeck, University of London. Infants/toddlers with Down syndrome learn things at a slower pace and fall more and more behind typically developing children as they get older. From early on, they particularly struggle to develop their major motor skills (e.g., sitting, crawling, walking). As they reach their second year of life, they start showing particular difficulties with expressive language. However, large differences exist across children with Down syndrome. The focus is now on trying to understand why some children with Down syndrome do relatively well compared to others, as this may provide important insights for early interventions.

Rifdat Aoidi presented the work of Victor Tybulewicz lab at The Francis Crick Institute. 50% of babies with Down Syndrome are born with heart diseases also called “hole in the heart”. Because the human genome share 92% of their genes with the mouse, researchers have been using the mouse to model human conditions. The Tybulewicz lab has generated mouse models that reproduce the heart diseases observed in babies with Down syndrome and we are using these mice to understand the heart development in Down Syndrome and to identify the gene(s) on chromosome 21, that cause heart diseases.

Carla, one of the researchers working with adults with Down syndrome at King's College London (KCL), spoke about some of the team's findings looking at cross-sectional changes in people across the lifespan. The team at KCL found that people's cognitive abilities decreased as they got older, with earlier changes seen in memory and attention compared to changes in planning, coordination, verbal abilities, and everyday abilities. The team are now looking to confirm these findings in longitudinal studies. Carla also presented some data from adults and infants with Down syndrome which showed epilepsy, hypothyroidism, and cataracts all become more common as people get older.

At the end of the scientific talks, one of the people that participated in the studies, Harriet, shared her experience of living with Down syndrome. Harriet is 45, lives in her own supported living flat within bus-distance of her parents’ house, she works at a museum and a cinema, and she goes to church on Sundays. When asked about what she was proud of, she simply replied *‘everything’*.

For a few hours on world down syndrome day 2018, in the state-of-the-art Francis Crick Institute, scientists and people who have Down syndrome had the chance to chat and share each other’s everyday experiences. What was the purpose? For our participants: to feel closer to science and to understand the ways their participation can help us to solve seemingly impossible problems. To us scientists: to remind us that our ultimate goal lies beyond all the data, cells, numbers and pathways to the people in that room, and the possibility that we can positively affect their lives.

The LonDownS Consoritum

The members of the LonDownS Consortium are: Andre Strydom, Frances Wiseman, Elizabeth Fisher, Victor Tybulewicz, Michael Thomas, Dean Nizetic and John Hardy.

References

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LonDownS Consortium Website**:** https://www.ucl.ac.uk/london-down-syndrome-consortium

Sinai, A., Mokrysz, C., Bernal, J., Bohnen, I., Bonell, S., Courtenay, K., . . . Strydom, A. (2017, 12 19). Predictors of Age of Diagnosis and Survival of Alzheimer’s Disease in Down Syndrome. (E. Head, Ed.) Journal of Alzheimer's Disease, 61(2), 717-728. https://www.ncbi.nlm.nih.gov/pmc/articles/PMC6004911/pdf/jad-61-jad170624.pdf

T21RS is the first, and only, non-profit scientific organization of researchers studying Down syndrome, founded to promote research, apply new scientific knowledge to develop improved treatments and cures, and to explain (recent) findings to the general public. More information? www.T21RS.org