



OBITUARY

The Joy of preimplantation genetic testing



Joy Dorothy Ann Delhanty (1937–2021), an original human cytogeneticist, cancer geneticist and pioneer of preimplantation genetic testing (PGT) died in

October 2021. Her last words apparently were, 'make sure you get this published'.

Joy joined University College London in 1956, where she stayed until the end of her career. A student of Lionel Penrose, she is credited with discovering the first human triploid. Subsequent population studies have established that triploidy is one of the leading causes of first trimester miscarriage and that screening for it and other types of chromosome abnormality in pregnancy loss provides comfort to many families. She also discovered the first reported case of heritable Down Syndrome. Caused by a Robertsonian translocation in a carrier parent, it is now a key part of genetic counselling, and similar subsequent studies in non-human animals have ramifications for farming and food production. Joy's group also identified novel mutations in families affected by genetic changes that arise in an inherited form of colorectal cancer (adenomatous polyposis coli) and discovered that the severity of the disease could be related back to the specific inherited mutation in the family. The importance of these findings proved to be that genetic testing could be used to counsel family members: those that did not carry the mutation had a much-reduced risk of developing colorectal cancer, whereas, if the mutation was identified in a family member, a tailored treatment plan could be designed.

Joy pioneered the use of fluorescence in-situ hybridization (FISH) for PGT,

performing the first cytogenetic clinical uses of PGT. Perhaps her greatest contribution to science is her body of ground-breaking work that informs us that the human embryo is cytogenetically not as the textbooks would have us believe. A uniformly diploid number of $2n = 46$ chromosomes in every cell, particularly at cleavage stage, is not the norm. On the contrary, early humans are karyotypically complex, fluid and dynamic.

It was sometimes said that 'Joy' was perhaps not the most appropriate moniker for someone who was so apparently serious, considered and quiet. In my opinion, this is unfair criticism. Joy was deep, there is no doubt.

Silences between the sentences of a conversation were commonplace and, when on the phone, could actually be quite disconcerting. But that was just Joy, invariably she would be thinking and considering her next response, and she did not waste words. She was nonetheless passionate about her science, would fiercely support her students and staff and created a powerful environment of discovery and learning in her laboratory. I personally remember the 'joy' in the Galton Laboratory (room 208) that was probably a direct result of Joy's management methods. I remember the Galton people that became lifelong friends and leaders in the field of PGT in their own right: Dagan Wells, Sioban Sen Gupta, Alan Thornhill, Joyce Harper, to name but a few. I remember the impressive collaborative colleagues such as Alan Handyside and Robert Winston and I am particularly grateful for the journey of discovery upon which she sent me.

Joy was rarely demonstrative, but always managed to create a buzz around her. I think that she actually looked on with some bemusement at the dynamic environment for which she was

responsible. Children rarely appreciate how lucky they are and the same applies to PhD students. But Joy let us make our own mistakes, picking up the pieces when necessary. She managed to get us around the world (quite literally in my case) to speak about our research at conferences. She got us published and publicised. I also felt the enormous pride of being asked back to UCL by her to be an external examiner both for MSc and PhD students. Such is her legacy, not only her academic 'children' but also the 'grandchildren' (students of her students), a phrase that, by the way she didn't care for. There are even a few great-grandchildren out there now. We will surely miss her.

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