

Julia Bell (1879–1979)

Julia Bell worked in twentieth-century Britain, discovered Fragile X Syndrome, and helped find heritable elements of other developmental and genetic disorders. Bell also wrote much of the five volume *Treasury of Human Inheritance*, a collection about genetics and genetic disorders. Bell researched until late in life, authoring an original research article on the effects of the rubella virus on fetal development (Congenital Rubella Syndrome) at the age of 80.

Bell was born 28 January 1879 in Sherwood, Nottinghamshire, England, to Katherine Thomas Heap and James Bell, a printer. Bell was born into a large family, the tenth of fourteen children. From 1898 until 1901 she studied mathematics at Girton College, the women's college at Cambridge University, in Cambridge, UK. Girton was then the only option for women to study at Cambridge, though the university didn't grant degrees to women until 1948. Bell eventually received her degree, but she couldn't complete her final examinations due to illness. Bell attained a Masters of Arts degree in Mathematics in 1907 from Trinity College in Dublin, Ireland.

Upon graduation from Trinity, Bell worked as an assistant to Karl Pearson, the director of the Galton Laboratory at the University College in London, a part of the University of London, UK. While working there, Bell pursued her medical degree. In 1914, Bell attended the London School of Medicine for Women, part of the University of London, where she received her medical degree in 1920. She then began her work on volumes two, four and five of the *Treasury of Human Inheritance*, a five-volume set of books that catalogued and analyzed genetic disorders. Bell worked on the *Treasury* for nearly five decades, and she maintained a relationship with the Galton Laboratory for most of her life.

While writing the *Treasury*, Bell investigated the method of transmission of Huntington's disease. She predicted that genetic markers could identify carriers of the disease even in the absence of symptoms, a prediction confirmed by data. Researching with John Burdon Sanderson Haldane, Bell also found a hereditary connection between colorblindness and hemophilia. The pair jointly published in 1937 a paper "The Linkage between the Genes for Colour-blindness and Haemophilia in Man" outlining the connection between the two genes. The genes that cause hemophilia and colorblindness were determined to be on the same chromosome, and this discovery facilitated research into areas such as chromosome mapping.

In 1943 Bell and James Purdon Martin discovered a form of sex-linked mental retardation that is inherited from the X-chromosome of a mother carrying the trait or an affected father (X-linked). The syndrome was initially named Martin-Bell syndrome after its discoverers, but the name was changed to Fragile X syndrome. In their paper "A Pedigree of Mental Defect Showing Sex Linkage," Bell and Martin discuss their study of a family who had taken an infant exhibiting symptoms of mental retardation to the National Hospital in London, UK. This family had a total of eleven males across two generations who exhibited symptoms of mental retardation. After interviewing affected individuals and detailing the family's history, Martin and Bell suggested that the condition was sex linked, heritable, and caused specific sections of the brain to develop improperly. Because the patients had difficulty with speech, the two researchers hypothesized that the pre-frontal cortex was the affected area.

Later in her career Bell remained active in research, publishing research on Congenital Rubella Syndrome in 1959. Bell collected information on the conditions of 712 infants whose mothers had contracted rubella during various periods during their pregnancy. She outlined a possible connection between a mother contracting rubella early in the pregnancy, before twelve weeks, and the fetus developing deafness, cataracts, or congenital heart disease. Although Bell's results were not

definitive, later studies supported her results. Bell's work helped lay the groundwork for the emerging field of teratology, which is the study of the causes and processes of abnormal development.

Bell remained involved with the Galton Laboratory in some capacity until 1965, when she retired at the age of 86. Bell never married, and lived alone until she entered a supervised care facility at the age of 97. When she was 100, Julia Bell died in St. George's Nursing Home in Westminster, London.

Sources

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