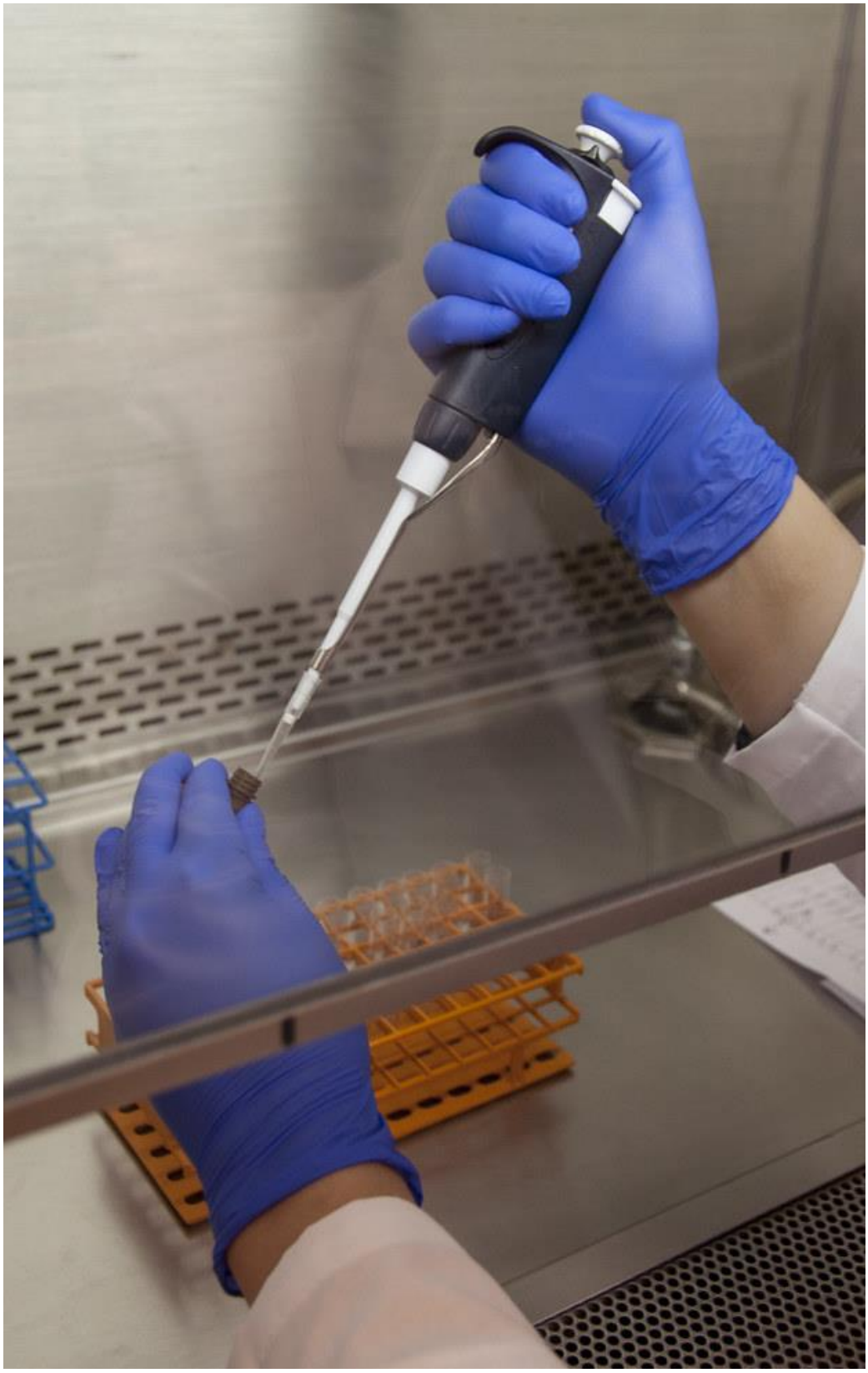
Study by DBT-InStem researchers offers newer insights into ASD/IDs

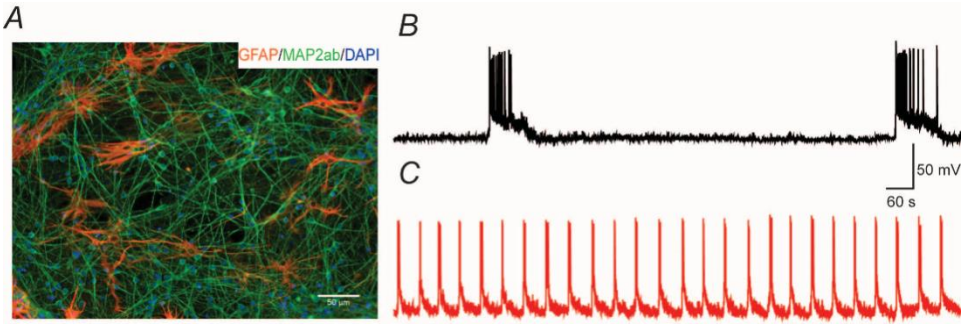


New Delhi, July 16: Autism Spectrum Disorders and Intellectual Disabilities (ASD/IDs) represent a major and growing public health challenge. Bengaluru-based Centre for Neurodevelopmental Synaptopathies (CNS) studies the pathophysiology of ASD/IDs. The Centre is supported by the Department of Biotechnology (DBT). It is a partnership between inSem and NCBS in Bangalore and the University of Edinburgh, UK.

A major effort at the CNS has been on accelerating the discovery of effective therapeutics for these debilitating brain disorders. Gene editing technologies offer an unprecedented opportunity for the creation of in vitro human based systems that model aspects of ASDs. Hence, CNS has established human induced pluripotent stem cell (iPSC)-based in vitro assays for discovering cellular mechanisms underlying ASDs.

A recent work led by Professors Sumantra Chattarji, David Wiyllie and Sidharthan Chandran in the CNS Programme has provided new insights into neural defects caused by Fragile X Syndrome (FXS) – the leading genetic cause of ASD/IDs. FXS is estimated to affect 1 in 5,000 men and 1 in 4,000-6,000 women worldwide. Rates of FXS have been calculated to be similar in India.

The work also throws light on the aberrant electrical activity patterns in human cortical neurons caused by FXS (see figure). Further, it identifies specific molecular mechanisms underlying these abnormalities, which can be reversed using pharmacological manipulations. Together, these observations offer new potential therapeutic targets that can be assessed in pre-clinical platforms and eventually lead to more efficient translation to the clinic.



The research findings of this work was recently published in `The Molecular Autism' Journal titled as 'Cortical neurons derived from human pluripotent stem cells lacking FMRP display altered spontaneous firing patterns.'

The paper was authored by Das Sharma S., Pal R., Reddy B.K., Selvaraj B.T, Raj N., Samaga K.K., Srinivasan D.J., Ornelas L., Sareen D., Livesey M.R., Bassell G.J., Svendsen C.N., Kind P.C., Chandran S., Chattarji S. and Wyllie D.J.A.

Links:

Centre for Neurodevelopmental Synaptopathies (CNS): **https://www.instem.res.in/bddm/cns**

Research paper: https://molecularautism.biomedcentral.com/articles/10.1186/s13229-020-00351-4