



THE HOSPITAL FOR
SICK CHILDREN



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Re: Support letter for Francesca Mari

I am delighted to support the grant application by Francesca Mari to the "Italian Ministry of Education, University and Research" with the title "X linked Intellectual Disability (XLID): development of human cellular models and identification of new candidate genes". We have ongoing collaboration with the group in Siena which has already resulted in a joint publication in the *European Journal of Human Genetics*. This paper is entitled "iPS cells to model CDKL5-related disorders" (by Amenduni M, De Filippis R, Cheung AY, Disciglio V, Epistolato MC, Ariani F, Mari F, Mencarelli MA, Hayek Y, Renieri A, Ellis J, Meloni I. *Eur J Hum Genet*. 2011 Dec;19(12):1246-55).

It will be a pleasure to extend our current collaboration by supporting Francesca Mari in her project with my expertise on induced Pluripotent Stem (iPS) cells. To this end, I will provide her with the MECP2+/R306C iPS cell line that we developed in our laboratory (Hotta et al, *Nature Methods* 2009 and Hotta et al *Nature Protocols* 2009). This line is now being distributed by the Coriell Collection (product GM 23298*A) demonstrating independent validation of its high quality.

I am confident that this approach will provide important insight into the molecular pathways involved in Rett syndrome and will facilitate drug discovery for this tragic childhood disorder.

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