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Dipartimento di Biotecnologie Mediche

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To the Rett Syndrome Europe President
Thomas Bertrand
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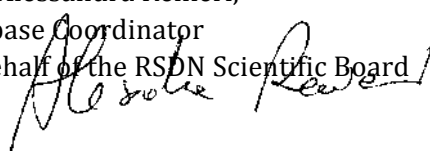
Dear RSE President,

the "Rett syndrome database network" (RSDN) was established in 2008 thanks to the EuroRett grant funded by E-rare (Coordinator Laurent Villard). It was conceived to collect clinical and genetic data of a huge number of Rett patients from different European countries and from countries outside Europe, in a standardized and easily comparable manner. The main purposes of this networked database are: i- epidemiological studies; ii- genotype-phenotype correlations, iii-modifier genes identification, iv- patients selection for clinical trials. At the moment it includes more than 2000 patients from 13 different countries.

In 2012, a first article on the RSDN was published (Rett networked database: an integrated clinical and genetic network of Rett syndrome databases. Grillo E et al. Hum Mutat. 2012. 33:1031-6), and a second article is planned by the end of 2015. For this purpose, in the second half of 2014 and in the first months of 2015, all the involved centres made a huge effort for patients' follow-up, for the update and the completion of clinical and molecular data insertion. To ensure that this effort by the referring clinicians can continue, we need to improve the software in a more user friendly manner in order to reduce the average time for filling patient information for each contributor. In addition, in order to analyse the inserted data, there is the need to improve the data mining system of the database. An additional implementation will be also necessary for patients' selection for clinical trials.

For all the above-described aims, we thus ask you to promote Database funding among the Parents' National Associations. In the past years we had requested a grant of 5,000 Euro to the largest Associations, and a lesser amount (not less than 1,000 Euro) to the smaller ones. With the obtained support, we could not be able to cover the entire costs of the Database Administrator, as previously shared during the periodic Scientific Board Conference Calls. Furthermore, in 2014 we did not receive any funds from the Associations. Thus, we have not been able to perform any improvement to the database and this last year, as already asserted, was used to ameliorate patients' data thanks to a great effort of each involved clinician. We ask, therefore, to help us getting funds for 2014 and 2015 with which we could cover the backlog costs and make the improvements described above.

Thanks for your collaboration
Best Regards

Prof. Alessandra Renieri,
Database Coordinator
On behalf of the RSDN Scientific Board


cc Laurent Villard,
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