



Kind Doctor,

we are contacting the families with children carriers of RING14 syndrome and of the Deletion of the Chromosome 14, to involve them in the project of genetic research that you will find in enclosure.

In these first 2 years since our inception our efforts have been focused on two issues:

- the first one has been to find all the families with child carriers of the RING14 syndrome or of a Deletion of chromosome 14,
- the second has been to collect money for us to be able to finance research.

Thanks to the collaboration of so many friends, to good relationships with other international associations (we thank especially: Unique, CDO, North, Mum's, Chromosome Help Station, Valentin Apac, 11Q network and Leona, Gen Polonia) and to the visibility of our web site we have succeeded in coming into contact with about sixty families in the world.

The collaboration that we ask to the families is to send samples of your blood and of your child's blood to the laboratory in Rome where it will be analyzed.

This is a first, very important, step, followed by, we hope from next autumn, other correlated projects involving the study of the neurological symptomatology and the creation and maintenance of a database of the syndrome. We will give you information about these second levels of the project over the next few months.

As a parents' association, our objectives are to make a start on genetic research that can give answers for the long time term, and, with the start of the medical projects next autumn, to give in brief terms, answers to questions about therapies and rehabilitation in order to improve the health and development of our children as soon as possible.

We explain to you in detail procedures for collection of blood samples and how to send them. If you have any doubts or problems, please contact Prof. Zollino in the Centre for Medical Genetics of the Università Cattolica Sacro Cuore in Rome ITALY to the followings address: tel +39 -06-30154927 or +39 -06-30154606 or +39-06-3050031
e-mail: mzollino@rm.unicatt.it

We confide in your collaboration to help the family to participate in this important project and we stay to your disposition for any further explanation about it

We have sent the following protocol to the parents for the procedure regarding the dispatch of the blood:

- 1) You have to make sure that the hospital or doctor takes the blood sample early in the week (from Monday to Wednesday). Fasting is not obligatory.
- 2) Blood will be inserted in 2 Sterile test-tubes and samples must be taken from both parents and from the child with the following requirements and to be shipped at Temperature Environment

- 3 mls in EDTA (emocromo test-tubes) and
- 7 mls in SODIUM HEPARIN (in the absence of specific test-tubes, the specimen can be performed in heparinized syringe and then transferred to a test-tube)

3) the test-tubes must be sealed with adhesive tape and inserted into 2 sheets of polystyrene with adhesive tape and finally inserted into a padded envelope to make a total of 3 packs. Before taking the sample you have to buy your packaging and have it ready because after sampling you MUST send the blood that same morning. You must keep in mind you will send 6 test-tubes (2 for mother, 2 for dad and 2 for child) therefore you don't need a big pack: the padded envelope can have dimensions of around 23 cm x 16 cm.

4) The same morning you have to go to the nearest shipping office (DHL, TRACO, EXECUTIVE.....)to ship the the consignment. Make sure you have the address to send it to with you.

5) At the shipping office you will fill out a consignment form with your address as sender and the following address of the recipient:

Prof.ssa Marcella Zollino
Istituto di Genetica Medica Università Cattolica Sacro Cuore
L.go F. Vito, 1
00168 Roma ITALY

you must also specify the following telephone numbers of Prof. Zollino:

tel +39-06-30154927 or +39-06-30154606 or +39-06-3050031

6) You must specify that it contains non-infectious blood samples.

7) send us an e- mail to tell us about the consignment so that we can tell the university and check that it arrives.

It is our intention to phone each of you to get to know you better and to be able to answer any further questions you might have.

We will pick up the cost of the molecular analysis but you have to pay the cost of the consignment because we don't have the possibility to pay from here. Anyway if you prefer you can send us by post or by fax the amount you have paid and we will refund it to you.

I am happy to explain any of this more to you if you have any questions. Thank you very much for collaborating with us.

Best regards

Stefania Azzali
 President RING14

Associazione internazionale RING14 Onlus
per la ricerca sulle malattie neurogenetiche rare

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