

Prof. Marie-Odile Rethoré Her Patients and Research – *a Life* –



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The Trisomy 21 Research Society (T21RS) awarded an honorary membership to professor Marie-Odile Rethoré. Marie-Odile Rethoré (Paris, France) is 89 years old and a well-known clinician who devoted all her life to patients with Down syndrome during her clinics.

During her life, Marie Odile Rethoré took care of people with Down syndrome. Day after day (and sometimes night after day) she gave advice and support to family members and other caregivers. She initially worked with prof. Jérôme Lejeune in the Trousseau Hospital in Paris and continued at the Jérôme Lejeune Institute. She was full-time in charge of young children with intellectual disabilities, mostly Down syndrome, and followed her patients' life from birth until old age. She has more than 160 publications, of which almost 40 are on Down syndrome research. She provided trainings to caregivers, physicians and families. She is member of many well-known scientific academies, in particular the Academy of Medicine in Paris.

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Her Patients

Prof. Rethoré sees every patient as a unique one, an individual. To the parents, Rethoré used to say *“Don’t be afraid of speaking about your anxieties, your rebellion, your dismay, your feeling of guilt. Keep well in mind that no other parent is better prepared than you are to be the father or mother of this child.”*. The relationship she established with her patients at different ages, and during each visit is important: from young baby to old-age, she saw them going through childhood, puberty and adulthood and she continuously took care of them. She considered every new step compared to a previous visit, mentioning it to the parents with encouragement and counselling. She helped develop the Intelli’cure® program to facilitate training for parents. As Rethoré stated: *“Don’t hesitate in asking questions about the disease of your child. The day comes when the child decides he wants to become an individual of his own. By giving birth, his mother brings him into the world, offers him to the world.”*.

Promotion of the dignity of her patients is a recurring preoccupation. *“It is our duty, as adults, to make sure that, during his whole life, this world respect his dignity as a man in all the various aspects of his personality. Nothing should be done if, even if it means renouncing to projects, programs or interventions sometimes proposed with the best intentions in the world but which risk hiding the reality in order to satisfy someone else’s own desires.”*

Her research

Rethoré particularly devoted her research to patient’s phenotypes (physical characteristics) and karyotypes analysis, to the careful description of malformations and dysmorphisms and in particular to the dermatoglyphic (study of skin markings or patterns on fingers, hands, and feet, and its application). In parallel with better knowledge of the chromosomes, she contributed to a better knowledge of the human chromosomal pathologies. A lot of morbid entities were described firstly by her and particularities on

different chromosomes (chromosomes 3, 4, 9,11, 12,16,17, 19) and different particularities on chromosome 21 from 1970 to 1986. The technology to analyze karyotypes (analysis of the chromosomes within the cell) was really more difficult in that time. And, more deeply, the analysis of the detailed structure of the chromatids (part of the chromosome) let her describe different original chromosomal rearrangements (translocation, chromosomal insertion, juxta centromeric monosomy, centric fusion between homologous chromosomes). As for the co-discovery by Jérôme Lejeune and Marthe Gautier of the third chromosome 21 which occurred in 1958, one year following her Doctor of Medicine (MD) graduation (1957), each further Rethoré’s new publication *“authorize to explain each abnormality of the kids and to give parents some explanation concerning the new discovered hereditary material abnormality!”*. Moreover, in collaboration with different biochemistry department, she determined the specific localization of some enzymes and in particular on the chromosome 21 the Superoxide dismutase (1976), the phosphoribosyl glycinamide synthase and cystathionine beta synthase (1984). Some of those enzymes are always nowadays source of research by international teams. Many publications in international papers presented her research.

Clinical work and research

The last 20 years at the Jérôme Lejeune Institute in Paris, her daily work- in collaboration with Jérôme Lejeune until his death in 1994 – contributed to the knowledge about Down syndrome. She studied some different particularities of the Down syndrome population and associated diseases in particular malignant diseases (i.e. breast, brain and testicular tumors) and immunological disease such as Coeliac disease or gluten intolerance. She was an important contributor to the Onco defi® program exploring malignant diseases in patients with intellectual deficiencies. Her interest led her to work on clinical, psychological, biological and anatomy analysis and risk factors of aging in Down

syndrome patients. She continuously emphasized the fact that interconnection between daily clinical practice, strong relationship between the physician and the patient and with the patients' family or caregivers is an immense source of novel topics for research and publication. She always looks for some correlation between different children, even unrelated, some new

and previously unknown aspects to thereafter engage a discussion with her colleagues. Most of them will never forget her notices, always pertinent. This is again an essential quality for clinical research. *"Since many years I am learning my physician work in front of thousands of patients with intellectual deficit seen in clinics with their families"*

Summary by prof. Rethoré herself

"I am so admiring in front of these parents who day after day, tirelessly are going from professionals of the care to other professional of the care to find the good one or ones who will to spark the best of their child. I am dreaming of physician colleagues all viewing at the same time on the same child or adult to answer to the parent's request! "to take in charge of patients with intellectual deficiencies into a same approach! Don't be afraid" of loving your child, of adopting him every morning when starting a day that might be quite agitated or the monotonous repetition of every day preceding this one "Don't be afraid of being happy with him, of telling him so, of showing him."

Further reading

The 50th anniversary of the discovery of trisomy 21: the past, present, and future of research and treatment of Down syndrome. Mégarbané, A., Ravel, A., Mircher, C., Sturtz, F., Grattau, Y., Rethoré, M.O., Delabar, J.M., and Mobley, W.C. *Genetics in Medicine* 2009 11(9):611-616
<https://www.ncbi.nlm.nih.gov/pubmed/?term=10.1097%2FGIM.0b013e3181b2e34c>

