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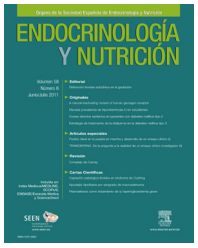

LETTERS TO THE EDITOR

Creating a European consortium to study GnRH deficiency (COST Action BM1105)☆

Creación de un consorcio europeo para el estudio de la deficiencia de GnRH (Acción COST BM1105)

Gonadotropin-releasing hormone deficiency (GnRHD) is a rare disease (prevalence, 1/10,000) characterized by the absence of puberty, infertility, and high psychosocial morbidity. GnRHD has a strong genetic component. Different approaches such as genetic studies (chromosomal aberrations), homozygosity mapping in inbred families, and candidate gene analysis have allowed for the reporting of 16 genes whose mutations cause the disease.[1–4] Advances in this field continue to be slow, reference centers are few, and a genetic cause is not found in a majority of patients (two-thirds). In order to develop a European register of patients with GnRHD and to create a network of clinicians, translational researchers, basic scientists, biocomputing specialists, and genetists to promote new discoveries in the field of human reproduction, an action project was presented to the European Cooperation in Science and Technology (COST) program which was approved in 2012.

COST is an initiative based on an intergovernmental agreement of European Union countries and other member countries (a total of 36 COST countries www.cost.eu/about_cost/cost_countries) started in 1971 to potentiate, coordinate, and disseminate the scientific and technological research activities of networks formed by COST countries by supporting cooperation and interaction between researchers. COST reflects the human dimension of science, helping researchers to share not only the results of their work, but also their objectives and methods. COST funds come from the Framework Program and are allocated to finance cooperation networks called COST Actions. Each COST Action receives a financial contribution (on average 75,000–130,000 € per year, per action), based on a four-year joint work program, to be used for scientific management and work group meetings, also including short scientific stays for young researchers.

COST is divided into nine domains or scientific-technical areas and a transdisciplinary domain. The COST secretariat is held by the General Secretariat of the Council of the European Union at Brussels, which emphasizes its intergovernmental nature.[5,6] Finally, COST is open to participation by other countries, thus also contributing to increased worldwide cooperation. Institutions from countries which are not members of COST may join an action after a case-by-case study with no need for formal agreements between governments or agencies.

In the COST BM1105 Action, entitled ''GnRH deficiency: elucidation of the neuroendocrine control of human reproduction'' and included in Domain 1 (Biomedicine and Molecular Biology), the clinical group will develop an anonymous computer database to include phenotypes and genotypes of at least 1000 patients with GnRHD, their families, and healthy relatives; it will prepare updated clinical guidelines for genetic counseling and the optimization of the clinical management of these patients; and it will provide information material and links to patient support groups. The group of genetists and biocomputing specialists will in turn assist in the use of the most advanced genetic technology and in data interpretation, with the aim of identifying one or more altered genes in each patient and/or 10 or more new genes causing the disease. The group of basic scientists will prioritize the candidate genes identified by whole exome sequencing and will explore the biologic functions of new genes in animal or cellular model systems. Finally, the education and training group will coordinate the training program for young researchers.

More than 100 participants from 25 countries, including Spain, have joined the COST BM1105 Action (www.cost.eu/domains_actions/bmbs/Actions/BM1105), which has already been cited in a scientific publication[7] and has created its own web site (www.gnrhnetwork.eu). We hope that this European consortium, of which the authors are part, will help to speed up scientific advance in the field of GnRHD, including the description of new biomarkers and individualized therapies for infertility, thus representing a significant clinical benefit for patients and their families.

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Comments on «Importance of ultrasound in a department of endocrinology»☆

Comentarios sobre «Relevancia de la ecografía en un servicio de endocrinología»

Sir,

I read with great satisfaction the editorial of your journal where I. Argüelles and S. Tofé proposed the wide use of neck ultrasound examination at endocrinology outpatient clinics.[1] The defense of the participation of endocrinologists in clinical procedures where they are most efficient is no less than the defense of health system users. This is something that our specialty has not always been able to accomplish.

The department where I work has been performing neck ultrasound examinations at outpatient clinics since 2009. Our experience has some unique characteristics that may be of interest for the readers of ENDOCRINOLOGÍA Y NUTRICIÓN. Some time ago it was decided to locate the ultrasound equipment at the clinic occupied by the resident specialist during the 12 months of his/her training. The resident uses the equipment, supervised by an appropriate associate physician, for the diagnosis of anatomic changes and thyroidectomy follow-up. According to our data, each resident performs a mean of 192 ultrasound examinations and 108 fine needle aspirations (FNAs) under supervision. This agrees with data previously reported by editorial authors[2] showing that ultrasound equipment located at the endocrinologist clinic eliminates waiting lists for ultrasound-guided puncture. The performance of ultrasound-guided FNA at the endocrinology clinic has not weakened our relationship with the pathology department, but has actually reinforced it as we have jointly developed a modification of the technique that significantly decreases the number of non-diagnostic punctures.[3] We encourage endocrinology departments to install ultrasound equipments at their clinics, and agree with the authors of the text on the need for the Spanish Society of Endocrinology and Nutrition to position itself regarding our role in the performance of neck ultrasound examination.

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