I feel honored to be invited to participate in this **World Orphan Drug Congress USA** today in Washington, D.C. I would like to compliment the moderator, Roberto Luiz D'Ávila, president of the Federal Medicine Council, Congressman Jean Wyllys de Matos Santos, and Otavio Nunes, Interfarma Director of Institutional Communication.

I have been very close to this theme of rare diseases and orphan drugs because of my interaction with organizations. By direct influence of the Baresi Institute, São Paulo, coordinated by Adriana de Abreu Magalhães and Marcelo Dias Higa, there have been discussions regarding the public policies and the required social awareness for the subject regarding persons with these diseases. In Brasilia, I have had many dialogues regarding rare diseases, for example, with Rogerio Barbosa Lima, President of the Maria Victoria Association.

Inspired by the Baresi Institute activities, I presented three legislative matters on rare diseases. The first is the Senate Bill No. 159, from April 2011, establishing the last day of February as the National Day of Rare Diseases. The proposition is in the Senate Education Committee, awaiting the Chairman to arrange an open hearing for the bill´s regulation. The second proposal, introduced in November 2011, is Bill No. 711, establishing the National Policy for the Protection of Rights of People with Rare Disease. The matter was distributed to be analyzed by the Committees of Social Affairs (CAS) and of Human Rights and Participative Legislation (CDH), the latter being terminative.

Thus, there will be no need to go through the Senate Plenary to be sent to the House of Representatives analysis. The PLS No. 711 is with the *rapporteur* appointed by CAS, Senator Paulo Bauer, since March 29th, 2012. The third matter is the Senate Bill No. 231, of August 2011, establishing the National Fund for Research of Rare and Neglected Diseases. The project was distributed for the regulation of three committees: Science and Technology, Social Affairs and Economic Affairs, being terminative in the last one. Yesterday, april 10th, the Bill of Law was approved in the Social Affairs Committee, with a favorable report of Senator Ana Amélia.

It is interesting to point out that a rare disease is a pathology that seldom occurs in the general population. To be considered rare, each specific disease can not affect more than a limited number of people in relation to entire population. In Europe, the definition for

rare disease is one that affects one out of two thousand citizens (European Community Regulation on Orphan Medicinal Products). In the U.S, rare diseases are those affecting fewer than 200,000 individuals. In Japan, the legal definition of a rare disease is one that affects fewer than 50,000 patients in the country, approximately one out of 2,500 people.

Individually, each of the pathologies seen as rare affects less than one out of two people, but it must be emphasized: there are more than five thousand rare diseases identified. The etiology of rare diseases is diverse: the vast majority of them are of genetic origin (80%). However, degenerative, autoimmune, infectious and oncological diseases can also originate them. Despite the difficulties caused by rare diseases, many people suffering from them have provided and are still providing great contributions to humanity. I can mention as examples, President John Fitzgerald Kennedy with Crohn's disease; physicist Stephen Hawking with amyotrophic lateral sclerosis, the musician Seal, with children lupus, actor Michael J. Fox, Huntington's disease, the Olympic medallist Doug Herland, with imperfecta osteogenesis; and aviator Jessica Cox, who, by a congenital disease, was born without arms...

The sequels caused by rare diseases are responsible for the rise of near 30% of the deficiencies, which can be physical, hearing, visual, cognitive, behavioral, or multiple, depending on each pathology. The difficulty in medical treatment starts with the lack of a national mapping of these people. The low incidence of rare diseases in Brazil, compared to other countries, leads to the suspicion that many cases are simply not diagnosed, especially in regions historically not included, as the North and Northeast.

International data show that infant mortality among people with rare diseases reaches 30% in developed countries. This percentage may be even higher in Brazil, since these children do not receive proper treatment. Concrete data would be a basis to develop a coherent approach to the needs of this segment of the population. recent studies carried out by Eurordis – an organization that assists people with rare diseases in the EU – compared data from eight rare diseases in 17 European countries in a universe of 6,000 patients and families, showing that 25% of the patients surveyed waited five to 30 years between the appearance of the initial symptoms and final diagnosis. It is estimated that in Brazil, the difficulty of diagnosis and treatment are even higher, due to several factors including "The 10/90 Gap" phenomenon: according to the Global Forum for Health Research, less than ten percent of global spending on health research is dedicated to diseases and morbid conditions, representing ninety percent of the global burden of disease.

This "market failure" is characterized by a situation in which the private sector invests almost exclusively in drugs for the rich and

developed world, where they will be salable and profitable. As a result of this paucity of resources allocated to health research related to diseases of poverty, the so-called "neglected diseases", virtually ignored in terms of drug development, proliferate. Neglected diseases such as dengue, Chagas disease, schistosomiasis, leprosy, leishmaniasis, filariasis, river blindness, malaria, trachoma and tuberculosis, among others, are associated with poverty, with poor living conditions and health inequities, that is, with unjust, unnecessary and avoidable inequality. And President Dilma wants to put an end to them.

Even though they are responsible for almost half of the disease burden in developing countries, investments in research and development have not traditionally prioritized this area. Brazil, fortunately, is an exception to this rule: it has been already considered a world leader in research on neglected diseases. The initial actions of the Ministry of Health in relation to neglected diseases were launched in 2003, with the first themed announcement of tuberculosis, followed by announcements of dengue in 2004, and leprosy in 2005. In 2006, the Program of Research and Development for Neglected Diseases was established and 82 studies were funded, with a total investment of U$ 11.3 million. In 2008, 58 projects were funded through joint investment with the Ministry of Science and Technology and administrative support of the National Scientific and Technological Development (CNPq) of U$ 8.5 million, for a total of U$ 19.5 million. In 2012, the Ministry of Health announced the creation of a Neglected Diseases Research Network, with a U$10 million funding.

These are just some examples of government initiatives in this area. Besides the open hearing in the Senate to be held on April, 24th, announced today by Minister Alexandre Padilha, of Health, the Official Newspaper published a public consultation for the elaboration of norms and guidelines for the treatment of rare and neglected diseases.

In the case of rare diseases, the situation is similar to the one experienced by the neglected diseases, once the decision of the pharmaceutical industry to invest in research and marketing of products is largely influenced by demand and, especially, by the potential market. Based on this reasoning, the industry concentrates its production in certain lines, withdrawing from the market the drugs of low consumption, used in rare diseases, with low financial return or price controlled by the Government – once the biggest purchaser – as occurs with certain endemic diseases.

Orphan drugs are medicine used for the diagnosis, prevention and treatment of rare diseases. For a drug to be considered orphan, epidemiological criteria (low prevalence or incidence of the disease in a given population) and economic criteria (presumption of non-

profitability of the drug) are used. Both the diagnosis and treatment are hampered by lack of knowledge about these diseases and specific treatment protocols. There is also a shortage of specialized professionals capable of understanding the implications of treatments in a body with special features. This leads to the aggravation of symptoms and sequels. Often, people with rare diseases or associations that gather them are the ones in charge of locating the latest studies on their pathology, having them translated and forwarding them to their doctors.

People with rare diseases face huge social difficulties, often with insurmountable barriers. Prejudice against some common physical symptoms of these disorders is common, as well as the paternalist view that considers these individuals as a burden to society as opposed to an integral part of it. Many end up socially isolated due to lack of adequate infrastructure to their specific needs in schools, universities, workplaces and recreation centers. The vast majority of people with rare diseases do not have access to the necessary conditions to reach their full potential.

In this sense, it is very important to emphasize the importance of caregivers, who may be far greater in number when the Citizen's Basic Income is fully instituted in our Nation, because people will be able to make a choice of taking care of their loved ones, or of any family member who might suffer from problems of this nature. Brazil is the first nation in the world where the National Congress has approved the institution of a Citizen's Basic Income: the right of everyone sharing, no matter origin, sex, race, civil status, social and economic condition, to participate in the wealth of the nation. It will be instituted step by step, under the executive criteria, starting with those most in need, such as the Bolsa Família Program does today.

To deepen the studies on this subject and to establish better citizenship conditions for people with rare diseases, we need to contemplate them in all discussions about their needs. Having this great goal in mind, I introduced the bill establishing the National Day – and not just World – of Rare Diseases, to be celebrated, as throughout the world, on the last day of February. Despite recent scientific and technological advances, infectious diseases continue to disproportionately affect the poor and marginalized.

Increased investment in neglected diseases in Brazil, given the successes already achieved, now requires the institution of more permanent financing, that also contemplate rare diseases. In this sense, the creation of the National Fund for Research in Rare and Neglected Diseases, proposed by Senate Bill 231 of 2012, could mean an important advance with a view to broadening the financing and, consequently, the scope of the research actions in this area. The proposed Fund will operate under the forms of support for grants or repayable loans. The sources of funding include donations, being

allowed those for research of specific disease; grants and aid from entities of any nature, including international organizations, and refund of the loan operations carried out by the Fund as a of reimbursable funding. Moreover, resources will have annual budget of U$ 25 million, as proposed in my bill.

I would like to congratulate all the members of this World Orphan Drugs USA Congress. This is a very positive initiative to bring people of many nations together to study and to search for solutions in the struggle to deal with rare diseases. We should all keep the hard work to ensure, to people with rare diseases, the Brazilian constitutional principle that guarantees the right to health for all.