



# MODEL OF QUALITY OF LIFE IN RARE DISEASES

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## SUMMARY

It is considered a rare disease that affects less than 5 per 10,000 inhabitants. The WHO considers that there are around 7000 RD that affect 7% of the population. In Spain it is estimated that there are around 3 million people affected and according to the SIER report of 2013, it is estimated that in the Region of Murcia 88583 people suffer some rare pathologies. In a study carried out by FEDER, it was detected that 70% of the affected population has support needs that are often not covered due to lack of information or knowledge, and 45% are not satisfied with the health care received.

The Association of Rare Diseases **D'Genes** was created on January 2008, 25<sup>th</sup> and declared Public Interest Entity by the Ministry of Interior on June 2012, 29<sup>th</sup>. It is composed of parents, families, professionals in all fields in order to create opportunities for exchange and fellowship among friends and people diagnosed with Rare Diseases (RD), and awareness of the public health problem posed by low-prevalence diseases. It belongs to FEDER, EURORDIS and ALIBER, and among its main objectives are:

- ✓ Undertake actions that improve the quality of life and life expectancy of the patients and their families.
- ✓ Develop measures to avoid the social isolation in which most parents and patients find themselves.
- ✓ Carry out activities of diffusion of the characteristics and peculiarities of the diseases categorized as rare.
- ✓ Create a space where patients, family members and professionals can coexist and communicate.
- ✓ Raise awareness on the public health issue that the Rare Diseases represent.



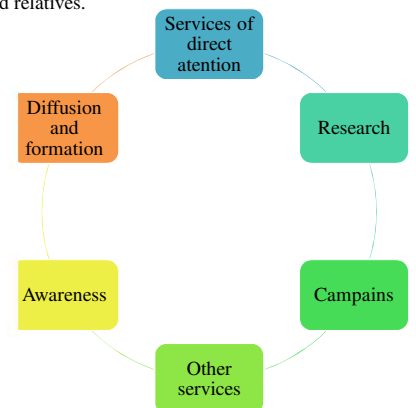
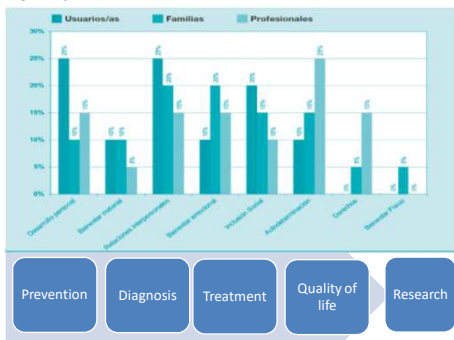
## D'GENES WORK MODEL

Taking into account our objectives and the characteristics that define the rare diseases and based on the Quality of Life Model of Shalock and Verdugo (2003), D'Genes has developed a working model from its beginning to the present, where it situates in the axis of his interventions to the affected and his family as the nucleus of his activity. It will be around this set from where the different coordinated work areas will be developed.

It is for this reason that the first indispensable work axis has been updated and quality training, from where we can know the latest advances in rare diseases (research, health policy, good practices, etc.) aimed at affected, family, professionals and students. The result has been the generation of new knowledge and the improvement of our direct attention services, which observes the affected from their capacities, taking into account the family and its community, as well as the rest of the environments so that inclusion is real and full. Likewise, the interventions constitute by themselves another source of knowledge, which allows that the treatments adjust more and more and better to the individual characteristics of each user. The Entity provides continuous, punctual, home and online services, in addition to developing programs that complete the global intervention.

Another important axis to complete the RD intervention is the dissemination and awareness of society, as a way to enhance inclusion and ensure an informed, plural and prepared community to serve all its citizens.

At present, D'Genes present a portfolio of activities that has the services of direct attention of Information and Orientation, Psychology, Physiotherapy, Speech Therapy, Cognitive Stimulation, Reflexology (all of them in their work centers or domiciliary modality), as well as Aquatic Therapy and Support in the Classroom (in the school itself). Online services such as Legal Advice and Psychological Support. In addition, since 2016 participates in the Pediatric Palliative Care program together with the University Hospital Virgen de la Arrixaca of Murcia. Parallel to the services, various leisure, leisure and free time workshops are given for children, adults and relatives. During the year 2017, 155 SIO cases were attended and 2120 sessions were offered.



In the lines of research, D'Genes participates in without diagnosis research project and, has developed the Educational Guide of X-Fragile Syndrome and is finishing a Psychological Support Guide to people with Rare Diseases. As for training, D'Genes has consolidated 10 years of work through its International Congress of Rare Diseases that in 2017 brought together more than. Specific work groups are also promoted for pathologies, creating spaces of coexistence and exchange of information where families and affected people.

## RESULTS

The results of our implantation generally entail a small measure of quality of life in the sample studied. Existing various surprises according to the informant is the affected person, his family member or the professional. For people with rare diseases, the most valued dimensions were those of personal development and interpersonal relationships. Complementary for the families were those of emotional well-being and interpersonal relationships. And finally, the professionals perceive that the most important thing in the quality of life would be the defense of their rights and self-denial of the person.

## CONCLUSIONS

The benefits of D'Genes work model are:

For patients: improvement of the quality of life, personalized and individual therapies where the person is contemplated from all their perspectives. Reduction of the feeling of loneliness.

For the family: reduction of feelings of loneliness and lack of information. Empowered family members who actively participate in decision making and offer greater security.

For society: promotion of a more informed, inclusive, adapted and prepared community to attend to diversity in all its forms.

Replicability of the model. D'Genes has been able to export its work formula through the different Centers that it has in the Region of Murcia.

## REFERENCES

1. Shalock y Verdugo (2003). Model of quality of life
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3. Plan integral de enfermedades raras de la región de Murcia.