

PRESS RELEASE



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Coordination and distribution of information on rare chromosome disorders is insufficient

Although recently more information became available on genes, heredity and chromosomes, the future of children with a rare chromosome disorder remains unclear. The Vereniging Samenwerkende Ouder- en Patiëntenorganisaties (VSOP, Dutch Genetic Patient Alliance) and the Chromosome Help-Station made an inventory of problems and wishes of people involved in rare chromosome disorders through 21 interviews with people and organisations in Denmark, France, Germany, Italy, The Netherlands, Norway, Poland and United Kingdom.

The study "European Rare Chromosomes United: a European orientation on people involved", published on February 11, 2004, concluded that there is little information available and that the coordination of (medical) care for people with rare chromosome disorders is suboptimal. Amongst others this is caused by the fact that the individual disorders are so different from each other and there is a huge heterogeneity between individual children with the same disorder. Although each individual disorder is rare as such, the total amount adds up to about 1 in 1000 births, who have a deletion or a duplication of part of a chromosome or a variant.

The problems of people with rare chromosome disorders are the rarity of the disorder, the lack of information on the disorder and the future perspective, the lack of contact with other people with the same problems, the huge influence on daily life and the fact that in most cases there is co-morbidity.

In the report the following recommendations are made:

More time for telling the diagnosis

After obtaining the diagnosis it is advised to give additional information in a second meeting with a geneticist. Also people have the possibility to ask questions. In the education of physicians more attention could be paid to communication skills needed to communicate these difficult messages.

Available medical information and experiences of people affected by should be better used.

Treating physicians and support groups could take care of assembling information and distributing the information along appropriate channels. Also the exchange of personal experience, which is felt by people involved as very valuable, could be facilitated through treating physicians (as already being done by outpatient clinics for rare chromosome disorders in The Netherlands) and through support groups.

More attention, care and supportive therapies provided throughout the entire life.

It is difficult to get optimal supportive care through the channels of regular care, partly because of the co-morbidity. A personal coordinator (key-worker) would be helpful to assist the parents in getting this care. For more specific care for rare disorders coordination on national (may be even international) level is desirable.

Stimulation of international contacts and support.

The rarity of the disorders asks for international cooperation. A restricted access to Internet and a lack of knowledge of English and scientific or medical terminology might hamper the access of people affected to the scarcely available information. It is advised that physicians help with translation of scientific information in laymen's language, a pool of translators could help to overcome language barriers.

The growing number of organisations for people with rare chromosome disorders, both at medical/scientific level as well as set up by parents of people affected, should join forces to improve the situation both at national as well as international level.

In the framework of a broader project planned the VSOP will investigate the possibilities of setting up expert centres / teams in the field of rare disorders. The Chromosome Help Station will initiate the discussion on his subject with support groups on a European level.

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For additional information <http://www.vsop.nl> en <http://www.chromosomehelpstation.com>

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