

Abstract

AISIA is a non-profit organization born in 2006 and made up of women, girls and parents directly affected by AIS and related conditions. In five years of activity we have been contacted by over 100 people, including affected women and their parents, spread all over Italy. We do not have a centre, people can contact us by email or by phone through our website www.sindromedimorris.org. In addition, every year we organize meetings for all the people who get in touch with us.

Our goals are: to offer support and information to all the affected people and families; to build a co-operative network which includes physicians and patients organizations; to become a point of reference for those who are interested in DSDs for personal or professional reasons.

Regione Lazio has recently approved and financed a project called IRIS, which consists of initiatives aimed at the diffusion of updated and deepened knowledge on DSDs. In particular, we will translate and distribute an important text published by ISNA in 2006, called "Clinical Guidelines for the Management of Sex Development in Childhood".

ABOUT US

AISIA is a non-profit organization made up of women, girls and parents directly affected by AIS. However, we have recently extended the scope of the group because an increasing number of PAIS diagnosis turn out to be something else: for example, 5-alpha-reductase deficiency. For this reason, we are now interested not only in AIS but also in related conditions, and we are in touch with parents of both girls and boys.

AISIA was born in 2006 inspired by AISSG UK, which has been active since 1996 and has got contacts with groups all over the world. Now we are in contact not only with the British group, but also with other European and non-European groups.

During our 5 years of activity, we have been contacted by over 100 people, including affected women and parents, spread all over Italy. We don't have a

centre, besides our web site www.sindromedimorris.org. People can get and stay in touch with us by e-mail or by phone.

Every year we organize an annual meeting open to all the people who contact us. We also organize psychological support and self-help meetings for affected girls and women.

OUR GOALS

1. To offer support and information to affected people and families.
2. To build and develop a co-operation network between physicians and patient organizations
3. To be a point of reference for those who are interested in DSD for personal or professional reasons.

1. To offer support and information to affected people and families

Meeting people who live our same experience is the key to dealing with a rare condition. This is why support groups become something like a second family, where people can talk, listen and share worries and experiences.

Sharing helps people to relieve the burden they bear, often in silence and solitude. This is true not only for those who are directly affected, but also and perhaps most importantly for their families. Parents of children with DSDs are often relieved when they meet affected adolescents and adults.

Meetings allow people to get in touch and to share experiences and feelings. As well, they offer precise and easily understandable pieces of information. Different aspects are discussed: not only the medical ones, but also social issues, affective and sexual relationships, infertility, adoption, etc.

Non-profit organizations which deals with DSDs gather reliable and updated information about the most recent medical researches and therapeutic options. They also collect data proceeding from the direct experiences of the affected people, especially regarding long term results of medical treatments and surgical interventions.

Information are subsequently made accessible to everyone through the web sites and through the publication of paper materials. To give an example, our organization has recently printed the Handbook for Parents in Italian, addressed to parents of children with DSDs. We have translated it from the original American one, which was developed through a joint effort of physicians, psychologists and ISNA (Intersex Society of North America).

2. To build and develop a co-operation network between physicians and organizations of patients.

Co-operation between patients and physicians can add to:

- Research projects (examples: psychological questionnaires, researches on osteoporosis and HRTs – Hormone Replacement Therapies)
- Testing the long-term results of the different therapeutic choices
- Production of scientific and non-scientific material
- Organization of meetings
- Building and developing a network of physicians and organizations interested in DSDs, both in Italy and outside it.

3. To be a point of reference for those who are interested in DSDs for personal or professional reasons

Non-profit organizations are a point of reference for people, information and experiences. In the specific case of rare conditions they perform a crucial role.

Physicians and patients are provided the opportunity to communicate and co-operate. Organization members often participate in medical meetings, and physicians participate in meetings and initiatives promoted by organizations.

It's a useful and functional way of co-operating for both sides. For this reason it should be encouraged.

On the one hand, organizations can help fulfill the patients' needs, while searching for updated information on research projects and new therapeutic options. On the other hand, physicians receive the opportunity to understand

patients' problems and difficulties, and to test the long-term effects of the different treatments they propose. This is helped by the familiar context in which the meetings take place, which allow people to express more freely their needs and concerns.

FUTURE PROJECTS

Recently Regione Lazio decided to sponsor a project of AISIA, called IRIS, which consists of initiatives aimed at the diffusion of updated and deepened knowledge addressed at everyone who is involved in a case of DSDs, including patients, families and physicians.

In particular, we would like to present you an informative project directed to physicians and health care workers who may have to handle DSD cases. It consists in the translation and distribution of an important text published by ISNA in 2006, called "Clinical Guidelines for the Management of Sex Development in Childhood". It also known as "Consortium on the Management of Disorders of Sex Development" and it is considered as one of the most important treatise on DSDs in the world. It is the result of the co-operation between physicians and patients.

The text aim to provide guidelines in order to support doctors and health-care workers involved in the management of DSD cases since childhood and throughout all the stages of their path, from the diagnosis and possible medical treatments, to the moment of communication and psychological support for the patients and their families.

The handbook presents the ideal setting in which a well-organized, multi-disciplinary, patient-centred team should work, in order to preserve the long-term well being of the patient. It also describe precisely which kind of specialists and health-care workers should compose such a team.

Two chapters are dedicated to the presentation of two protocols of the treatment of children diagnosed with DSDs: one dedicated to the newborns, the other to the children after the newborn period. Both describe in detail the steps to be taken (How and when to talk with the parents, Work stages of the team, etc) and deal with more specific problems, but always in a general way,

Some of the topics taken into consideration are:

- gender assignment;
- psychological support;
- timing of the hormone replacement therapy;
- behaviour norms and language to be adopted;
- necessity of telling the truth, and how to do it.

The text also include a chapter dedicated to the parents' most frequently asked questions, suggesting possible answers.

Finally, the handbook contains a list of resources (books, articles, websites, videos) that can be consulted and used in order to make the communication with patients and their families easier. Besides, each chapter include boxes related to important aspects to be considered.

We would like to stress that the project has been approved and financed by Regione Lazio. Therefore, the copies translated into Italian of the "Clinical Guidelines for the Management and of Sex Development in Childhood" will be initially distributed to all the major Italian centres of pediatric endocrinology and to the hospitals and local health authorities of Rome and Lazio.

Our organisation now consider a necessity to "provoke" in a constructive way the health care workers who handle cases of DSD, in order to build a team capable of pursuing a unique and common objective, of working together and involving the patients and their families in a continuous exchange of experience and information.

Indeed, it is crucial to work together in order to defeat fear and distrust. It is an objective that can be achieved only through the reduction of errors committed by doctors and health care workers, mainly because of the lack of information and expertise. In the past, patients have often seen their quality of life impaired in an irreversible way by such errors.

We want to make clear that AISIA does not want to take the place of the physicians. Nevertheless, five years after its birth our organisation has the duty of informing its members and everyone who asks for help about the physicians and health care workers who act according to the DSD guidelines recognised by the worldwide medical literature.

Therefore, we would like to see such an initiative welcomed and enhanced by all of you who are here today, and most of all by the representatives of the various pediatric endocrinology centres in Italy. We hope that they will commit themselves to back and follow these Guidelines and possibly that they will contribute to their improvement with their experiences.

In conclusion, we consider this project a crucial passage for our organisation. At the end of the project, in the second half of September, a final meeting will take place in Rome. In that occasion the activity of a year will be accounted, and the materials produced will be presented. We are also planning to assign a prize for the best dissertations on DSDs. This prize will be dedicated to the memory of Arianna Buttinelli, a member of our organization who gave an huge contribution to the birth and development of our organization, both on the human and intellectual level, and who sadly left us in September 2010. Organisations, patients, families and physicians will be invited.