

Clinical European study



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<http://www.dsd-life.eu>

Improving clinical care by exploring
needs and views of patients with various
conditions due to hormonal imbalances
of the gonads and adrenal glands

You are invited to take part in
this European study involving
15 medical centres in Europe.

Layout: Zentrale Mediendiensteleistungen, Charité - Universitätsmedizin Berlin



AIMS OF THE STUDY

Our **first aim** is to improve treatment and care of patients with various conditions due to hormonal imbalances of the gonads or adrenals.

This broad group includes e.g. Turner Syndrome, Klinefelter Syndrome, Mixed Gonadal Dysgenesis, Androgen Insensitivity Syndrome (AIS), XY and XX Gonadal Dysgenesis, Androgen Biosynthesis Defects (e.g. 5-Alpha-Reductase-2 and 17-Beta-HSD-3 Deficiency), Congenital Adrenal Hyperplasia (CAH) and Hypospadias. All of them are summarized under the medical umbrella term disorders of sex development (DSD). However, for various reasons some affected persons and interest groups do not agree to this term. In this study, these concerns will be taken into account. In particular, your suggestions for naming your conditions are requested.

Very little is known about the long-term outcome in this group in terms of long-term health, quality of life and psychological well-being. For this reason we want to evaluate the medical status, various aspects of well-being and general quality of life of patients, in order to develop European guidelines for treatment and care. The **second aim** of the project is to explore affected persons' satisfaction with their treatment and their views on how society, and medicine in particular, responds to their condition. The **third aim** of the study is to inform and educate medical professionals, nurseries, schools and the general public in Europe about needs and care of individuals with these rare conditions.

The priorities, in terms of medical and psychological issues, vary considerably between the different patient groups. The study will take account of these diverse concerns.

LONG-TERM CARE AND TREATMENT

Most persons affected by adrenal or gonadal hormonal imbalances need early and lifelong therapy with hydrocortisone, or therapies with testosterone or oestrogen starting at puberty. Some persons will have had reconstructive genital surgery in childhood or adulthood. Besides medical care, some persons may have had psychological counselling.

Although treatment and care may influence various aspects of well-being, the long-term effects of these interventions have not been studied in detail.

WHAT IS INVOLVED

We would like to investigate possible effects of the different therapies on health and general well-being. If you decide to participate, we need information about your medical history and medication. We also want to evaluate your current health with a medical examination and blood test, which will be performed by specialized endocrinologists, urologists or gynaecologists. Furthermore, we will ask you to fill in questionnaires to evaluate the various aspects of your well being.

YOUR VIEW

An important prerequisite to improve care is to know more about your personal experiences and ethical issues of treatment and care. These will also be investigated through the questionnaire.

COURSE OF THE STUDY

We would like to evaluate your health status through a medical exam, gynaecological/urological exam, blood test, bone scan, ultrasound and measurement of body composition. We would also like to learn about your experiences and your current situation through the questionnaires.

Your participation in one or more parts of the study is optional and your decision to participate or not has no consequences for your treatment. All personal information in the study is strictly confidential and personal data will not be transferred to anybody. Study data will be stored pseudonymized in an electronic database (dealt with under a different name or a number).

SUPPORT

If you have any questions on your health, the members of the study group will help to find answers or provide referrals. A psychologist will be at your disposal for non-medical concerns.

PARTICIPATION

If you want to participate in the study, please contact your local study centre and we will provide you with more details.

Travel costs will be refunded.