

Clinical European study on the outcome of hormone therapies, surgery and psychological support in disorders/variations of sex development (DSD)

B. Köhler, W. Arlt, C. Bouvattier, P. Chatelain, H. Claahsen-van der Grinten, P. Cohen-Kettenis, A. Nordenstrom, C. Pienkowski, A. Richter-Unruh, J. Slowikowska-Hilczer, C. Sultan, M. Szarras-Capnik, N. Reisch, U. Thyen, C. Wiesemann

*ECE, 16th Congress of Endocrinology
3.5.-7.5.2014 Wroclaw, Poland*

Funding 2012-16; <http://www.dsd-life.eu>

***Congenital conditions with incongruence
between the
chromosomal
gonadal
and phenotypic sex***

Actual medical DSD classification

Chicago consensus Statement on Management of Intersex Disorders, Pediatrics 2006



Sex chromosome DSD	XY DSD	XX DSD
<p>45,X0 and Variants (Turner Syndrome)</p> <p>47, XXY (Klinefelter Syndrome)</p> <p>45,X0/46,XY (mixed gonadal dysgenesis)</p> <p>46,XX/46,XY</p>	<p>Impaired testes development Complete XY gonadal dysgenesis Partial XY gonadal dysgenesis Ovotesticular DSD Gonadal regression</p> <p>Impaired testosterone synthesis e.g. LH-Receptor- defects, 3β-HSD-2 17β-HSD-3, 5α-reduktase-2 deficiency</p> <p>Impaired testosterone action Complete and partial androgen insensitiviy (CAIS, PAIS)</p> <p>AMH and AMHR defects</p> <p>Hypospadias, cloacal exstrophy</p>	<p>Impaired ovarian development Ovotesticular, testicular DSD</p> <p>Androgen excess:</p> <p>1. fetal: Congenital adrenal hyperplasia 21-Hydroxylase-, 11β-Hydroxylase-, 3β-HSD-deficiency</p> <p>2. feto-placental aromatase-, oxidoreductase-deficiency</p> <p>3. maternal: luteoma</p> <p>Dysgenesis of the uterus or vagina</p> <p>Cloacal exstrophy, MURCS</p>

Long-term hormone therapies

- ***Estrogen*** (*Turner syndrome, XY DSD, XX DSD*)
- ***Testosterone*** (*Klinefelter syndrome, XY DSD*)
- ***Cortisone, fludrocortisone*** (*CAH*)

surgery

- ***Vaginoplasty*** (*CAH, XY DSD*)
- ***Hypospadias repair*** (*XY DSD, hypospadias*)
- ***Orchidopexy*** (*XY DSD, hypospadias*)
- ***Gonadectomy*** (*XY DSD*)

Gender issues (*XY DSD, CAH, 45,X0/46,XY, 46,XY/46,XX*)

Impairment of

- *Quality of life (QoL)*
- *Psychological well-being*
- *Social Participation*
- *Sexual life*

- *Effects of hormone therapies on long-term health ?*
- *Controversies about timing and need of surgery*

- *Infertility*
- *Stigma*

- **Objective 1:**

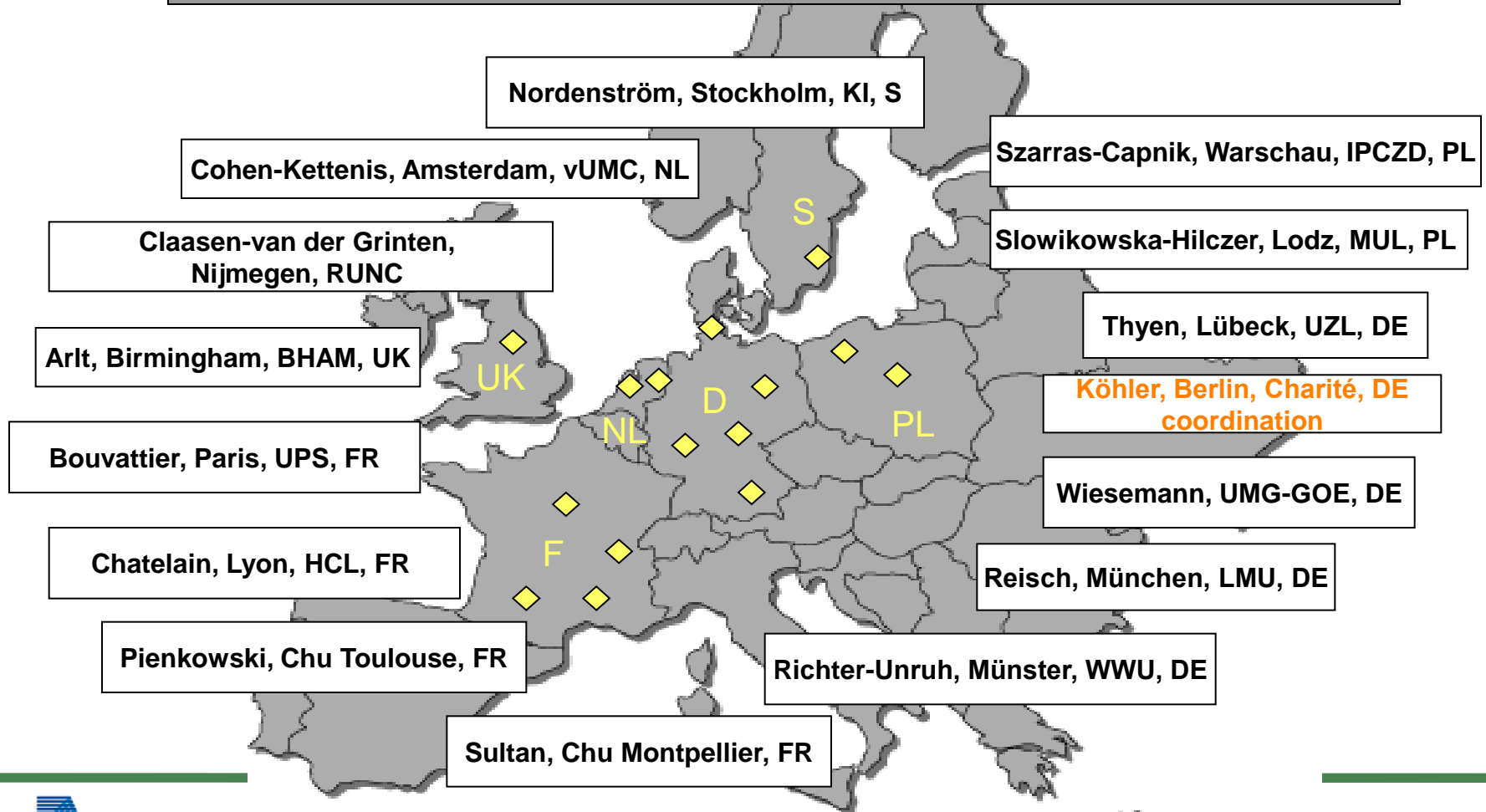
To improve clinical care of patients with disorders of sex development (DSD)

- **Objective 2:**

To develop clinical European guidelines for a better care of patients with DSD

14 Multidisciplinary Teams + Ethics

PeadEndo, Endocrinology, Psychology, QoL, Gyn, Urol, Andrology, Surgery



Evaluation of

- *QoL, psychological well-being and psychosocial adaption*
- *Long-term effects of hormone therapies on health*
- *Outcomes, need and appropriate age of surgery*
- *Patients' view on treatment and care*
- *Ethical issues*
- *Specific needs of patients with different cultural backgrounds*

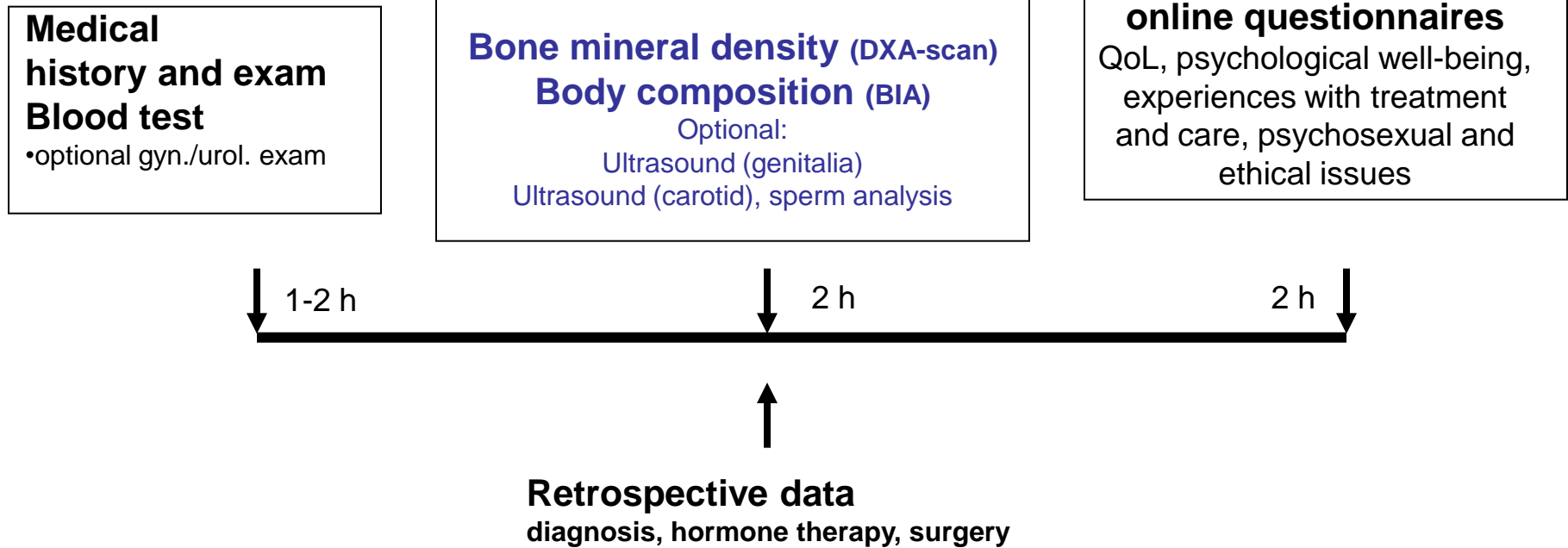
Four working groups

- *Psychology and QoL*
- *Medical issues*
- *Surgery and Gynaecology/Urology*
- *Ethics*

- ***Advice from patient support groups (CAH, XY DSD, Turner, Klinefelter syndrome)***

Participants \geq 16 years

Sex chromosome DSD	XY DSD	XX DSD
<p>45,X0 and Variants (Turner Syndrome)</p> <p>47, XXY (Klinefelter Syndrome)</p> <p>45,X0/47,XY (mixed gonadal dysgenesis)</p> <p>46,XX/46,XY</p> <p>500 patients</p>	<p>Impaired testes development Complete XY gonadal dysgenesis Partial XY gonadal dysgenesis Ovotesticular DSD</p> <p>Impaired testosterone synthesis e.g. LH-Receptor- defects, 3β-HSD-2 17β-HSD-3, 5α-reduktase-2 deficiency</p> <p>Impaired testosterone action Complete and partial androgen insensitiviy (CAIS, PAIS)</p> <p>Hypospadias</p> <p>500 patients</p>	<p>Impaired ovarian development Ovotesticular DSD</p> <p>Congenital adrenal hyperplasia 21-Hydroxylase-, 11β-Hydroxylase-, 3β-HSD-deficiency Oxidoreductase deficiency</p> <p>500 patients</p>



Qualitative interviews in a subset of participants

February 2014 - July 2015

- 1. Study centers and associated hospitals, clinics***
- 2. Patient support groups***
- 3. I-DSD registry***

- *QoL, psychological well-being, psychosocial adaption*
- *Influence of 1. sex assignment, 2. surgery, 3. hormone therapy and 4. psychological support on QoL, psychological well-being and psychosocial adaption*
- *Influence of co-founding factors such as metabolism, fertility, psychosexual development, social support, patients' view, patients' age and cultural influences*

Development of European guidelines for the management and care of DSD

Respecting the different needs

XY DSD

Turner syndrome

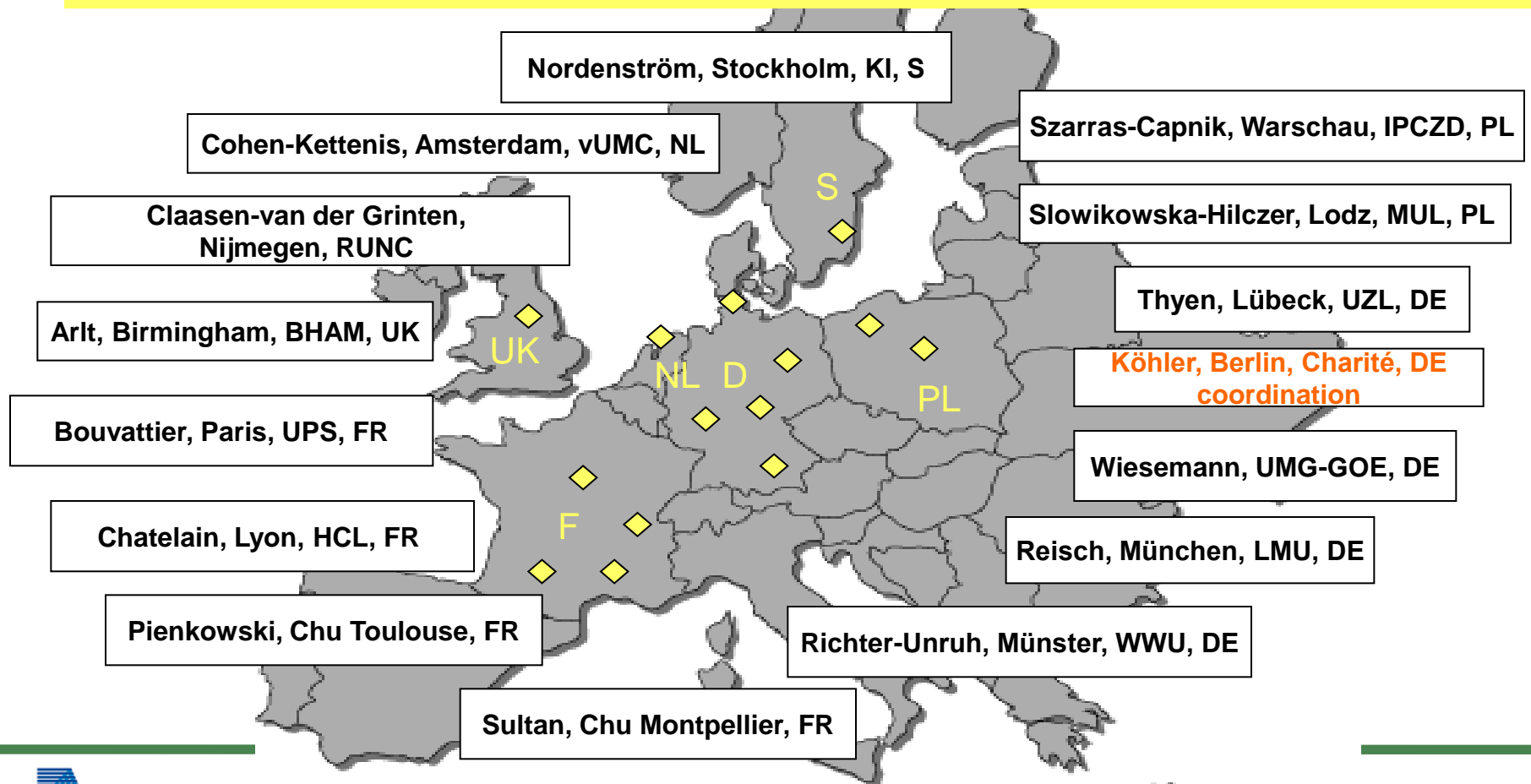
Klinefelter syndrome

CAH

Hypospadias

- *To promote general knowledge on the different diagnoses included in XY DSD, Turner and Klinefelter syndrome and CAH*
- *Information of educational and health care institutions*

We would be pleased if you could announce the study to your patients and promote participation in the study centers



Thank you on behalf of dsd-LIFE !



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