



RIKSHOSPITALET

University Hospital

Centre for Rare Disorders



A CENTRE TO TURN TO

Centre for Rare Disorders is an interdisciplinary, nationwide competence centre, which offers information, counselling and seminars on a selected range of rare disorders. The service is aimed at the appropriate patients and their families, and at professionals working with these disorders. Anyone may contact the centre, and a referral or advance appointment is not required.

INFORMATION

The centre obtains and coordinates specialist knowledge on, and experience with, designated rare disorders . This information is distributed via counselling, seminar activities, information booklets, videos and the Internet.

COUNSELLING

Centre for Rare Disorders offers counselling and guidance to patients and their families and to professionals via:

- phone and e-mail enquiries
- meetings at the centre or at the treating hospital
- meetings in the patient's local area and/or home environment

SEMINARS

Centre for Rare Disorders arranges seminars for patients, their families and professionals. The seminars normally last from 1 to 5 days, and are held at various locations throughout the country. They offer medical information related to certain specific diagnoses, and also address more general issues, such as everyday life in kindergarten/at school/at work, nutrition, physical activities, national health benefits, rights and privileges. An important element of these seminars is the sharing of experience.

KNOWLEDGE BASE BRIDGING THE GAP

Regional health services generally have little knowledge regarding rare disorders. As a result, competence transfer and information meetings held in the patients' local area are fundamental to our centre's activities. We strive to ensure that the transition from one stage of life to the next is planned well in advance, so that necessary adjustments can be implemented.

PATIENT PARTICIPATION

Centre for Rare Disorders collaborates with patients, their families and relevant patient organisations. The centre has a Centre Council where patients, represented by the patient organisations, and professionals make suggestions for the organisational development of the centre.

RESEARCH AND DEVELOPMENT

Research and development activities at the centre are run through collaboration between patient organisations and specialist environments at home and abroad. New international research and results from our centre's projects are reported to patients and relevant specialists.

The centre is a national undertaking and works on behalf of the Norwegian Directorate for Health Affairs. The centre is a division of Rikshospitalet University .

Links to Nordic diagnoses descriptions and contact information can be found at www.rarelink.no. If you need information on rare disorders, please call the service phone number **800 41 710** of the Norwegian Directorate for Health Affairs.

DIAGNOSES

Congenital anomalies

Anorectal anomalies

- Anal atresia
- Currarino triad

Bladder exstrophy/epispadias

Genital anomalies

- Adrenogenital syndrome (AGS), (CAH)
- Hermaphroditism – Pseudohermaphroditism
- Complete androgen insensitivity syndrome (CAIS)
- Partial androgen insensitivity syndrome (PAIS)
- Scrotal hypospadias

Oesophagus atresia

Cranofacial anomalies

Syndromatic craniosynostosis

- Apert syndrome
- Crouzon syndrome
- Pfeiffer syndrome
- Sæthre-Chotzen syndrome
- Muenke syndrome

Others

- Treacher Collins syndrome
- Hemifacial microsomia/ (OAVS)/Goldenhar
- Choanal atresia
- Cleidocranial dysplasia
- Microtia, anotia
- Cherubism
- Pierre Robin
- Freeman Sheldon Spectrum/ Whistling Face
- Trigonocephali

Congenital liver diseases

- Aagenaes syndrome
- Alagille syndrome
- Extrahepatic biliary atresia

Inborn errors of metabolism

- PKU – Phenylketonuria (Folling disease)
- Galactosaemia
- MSUD – Maple Syrup Urine Disease
- Methylmalonic acidemia
- Propionic acidemia
- Alport syndrome
- Fabry disease
- LCAT deficiency (Norum disease)

Congenital thrombocytopenia

Congenital haemophilia

- Afibrinogenemia
- Haemophilia A + B
- Owren's disease
- Factor VII deficiency
- von Willebrand's disease
- Hereditary hemorrhagic telangiectasia (HHT)

Primary immunodeficiencies

Hypogammaglobulinemia

- X-linked agammaglobulinemia
- CVID – Common Variable Immuno Deficiency
- IgG subclass deficiency
- Selective IgA deficiency
- Hyper-IgM syndrome

T-cell disease and combined B- and T-cell disorders

- SCID - Severe combined immunodeficiency
- Wiskott-Aldrich syndrom

Phagocytic deficiencies

- Chronic granulomatous disease (CGD)
- Interferon-gamma-receptor deficiency (INFRG)
- Leukocyte adhesion deficiency LAD
- Neutropenia

Complement deficiencies

Immunodeficiency associated with other syndromes

- Hyper-IgE syndrome
- Chronic mucocutaneous candidiasis CMC
- Autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy (APECED)
- Ivemark syndrome
- X-linked lymphoproliferative syndrome (Duncan's disease)

Congenital skin diseases

- Epidermolysis bullosa (EB)
- Ectodermal dysplasia (ED)
- Greither's syndrome
- Hydroa vacciniforme
- Ichthyosis
- Incontinentia pigmenti
- Keratosis
- Netherton syndrome
- Pachyonychia congenital

Other

- Mastocytosis

Hereditary eye and central nervous system disorders

- Laurence-Moon-Bardet-Biedl syndrome (LMBB)
- Huntington's disease
- Aniridia

CENTRE FOR RARE DISORDERS

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