

# ANGELMAN CENTER MUNICH

Initiative of the Children's Hospital & kbo-Kinderzentrum Munich Schwabing with Angelman e.V.



Zentrum für Kinder- und Jugendmedizin: Eine Kooperation der München Klinik und des Klinikums rechts der Isar der TUM



## WELL POSITIONED. WELL TAKEN CARE OF. WELL TREATED.

### Angelman Center Munich: Everything under one roof.

#### WHY?

Angelman syndrome (AS) is caused by a rare genetic defect on chromosome 15. The resulting lack of a single protein in brain cells leads to severe multiple disabilities. Characteristic features include frequent smiling, jerky movements (ataxia) and balance disorders which make it difficult to learn to crawl and walk. With increasing age, restricted movement frequently leads to foot malposition, hip maturation disorders and scoliosis. Often difficult-to-treat epilepsy, little or no speech development, extreme sleep disorders and challenging behavior, confront parents and attending pediatricians with the question of what measures are suitable for the overall picture.

#### OUR GOAL

All children affected aged 0 to 18 years should be regularly examined and treated at the Angelman Center Munich. Each child will undergo extensive initial and follow-up diagnostic procedures. Adult AS patients should be cared for at a clinic that works closely together with the AZM, so that an uncomplicated transition can take place.

#### OUR SERVICES

Pediatric neurologist Dr. Christine Makowski and the graduate psychologist Dr. Cornelia von Hagen lead and coordinate our multidisciplinary and competent team, in which specialists, therapists and consultants work hand in hand on topics of nutrition and supply of medical and technical aids. State of the art diagnostic procedures and treatments at the Angelman Center Munich are being carried out in line with international standards – this enables individual consultation with optimal therapy measures for each child. All costs for examinations and treatments are covered by statutory and/or private health insurance. The Children's Hospital and the social pediatric center kbo-Kinderzentrum Munich Schwabing are linked to the Technical University Munich by a cooperation agreement, where they work closely together with the Institute of Human Genetics. Here all diagnostic and imaging procedures are available to the Angelman Center Munich. Furthermore, the Angelman Center Munich cooperates with the University Hospital in Leipzig to update the Angelman patient register.

#### OUR CLAIM

The Angelman Center Munich wants to provide optimal care for all individuals affected by AS. It is supposed to be the point of contact that covers all diagnostic procedures and provides extensive consultation and assistance so that Angelman syndrome can be treated symptomatically and causally as soon as possible. The team members at the Angelman Center Munich are in regular exchange with international AS experts and have the expertise and unrestricted willingness to conduct clinical trials. The close and trusting cooperation with the parent association Angelman e.V. forms the basis for this.

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## DIAGNOSIS: ANGELMAN-SYNDROME

### this is how our treatment works ...

In order to obtain a clear picture of the respective manifestation of AS, a five-day in-patient stay should be planned for the first consultation at the Angelman Center Munich. In addition to physical examinations such as EEG, sonography of the organs and laboratory tests on blood and urine, detailed observations on the child's play and motor function are being carried out during the stay. This is complemented by collecting information via standardized questionnaires and test procedures on sleep and eating behavior, communication and language development as well as seizures and medication supply. The expert team of the Angelman Center Munich includes specialists from the fields of pediatric neurology, social pediatrics, child psychology, nutrition sciences, human genetics, pediatric orthopedics, speech therapy, physiotherapy, and therapeutic pedagogy. In order to broaden our findings, music therapy and neurofeedback will also be included in the consultations.



After evaluating the test results, a detailed consultation with specific proposals on treatment and preventive measures follows. Consultations should take place at regular intervals to ensure a differentiated follow-up examination. By analyzing all data collected at the Angelman center, an important contribution is made to the general course (natural history) of Angelman syndrome, which is of great importance for clinical studies on symptomatic and possibly causal treatments of AS. Studies with synthetic CBD for the treatment of epilepsy are already being conducted at the Angelman Center Munich. The Center is also participating in Ovid's Phase 3 NEPTUNE trial (OV101-Gaboxadol).

### In good hands ...



**Dr. Christine Makowski**  
Oberärztin München Klinik Schwabing  
Specialist in Pediatrics and Adolescent  
Medicine  
Focus on Neuropediatrics

“As a scientist and doctor, it is very exciting for me to help shape a new era of medical history through innovative therapies, thus decisively helping my patients. The cooperation with research-based pharmaceutical companies gives me the opportunity to quickly apply new treatment options clinically and enable individuals affected by AS to receive optimal therapy.”



*In-depth expertise can only be established with a large number of patients undergoing the individual examinations and interviews at the Angelman Center Munich. In this way physiological interactions can be identified and new research approaches be developed. Systematic analysis is also the prerequisite for adequate planning of therapy measures and intervention approaches. Cooperation with international experts enables a broad exchange of information making sure that current findings can be implemented immediately at our Center. During seminars and workshops, parents are being informed of treatment standards and current developments at regular intervals.“*



**PD Dr. Cornelia von Hagen**  
Dipl.-Psychologin  
Projektleiterin kbo-Kinderzentrum München Schwabing  
Wissenschaftliche Mitarbeiterin TU München



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Zuverlässig an Ihrer Seite

## TESTING at the Angelman Center

### Medical field:

- Genetics
- Standardized anamnesis
- Sonographic studies
- EEG
- Laboratory testing
- Radiological examination
- Rating severity AS (communication, sleep)
- Questionnaire Sleep Habits
- Seizure and Drug Questionnaire
- Questionnaire on nutrition and excretion
- MRI if required
- Intrathecal drug administration is standard at our university clinic

### Orthopedic field:

- Detailed orthopedic examination
- Radiological controls
- Treatment advice
- Adaption of aids (inserts, orthoses, Dynamic GPS soft-orthosis, storage elements, corset)

### Orthopedagogical field:

- Developmental milestones
- Questionnaire from the Institute of early childhood pedagogy
- Rating of AS severity (behavior)

### Nutritional advice:

- Ketogenic Diet
- Low Glycemic Index Diet

## MANAGEMENT

### Dr. Christine Makowski

Specialist in Pediatrics and Adolescent Medicine  
Focus on Neuropediatrics

### Priorities:

- Neuropediatrics
- Epileptologies
- Neurometabolic diseases
- Mitochondriopathien

### Memberships

- Scientific Advisory Board at Angelman e.V.
- German Society of Neuropediatrics
- German Society of Clinical Neurophysiology
- German Society of Epileptics
- German Society for Pediatrics and Adolescent Medicine

### Neurofeedback:

- EEG-Neurofeedback
- Hämoenzephalographie
- Quantitative EEG

### Physiotherapeutic field:

- Gross Motor Function Classification Scale
- Rating severity AS (motor skills)

### Instrumental gait analysis

- 3D analysis of gait patterns
- measurement of muscle activation
- Measurement of joint loads
- Pedobarography (foot pressure distribution)
- Special video diagnostics

### Psychological field:

- Bayley Scales of Infant and Toddler Development
- Vineland Social Maturity Scale
- Checkliste for deviant behavior
- Questionnaire on quality of life
- Questionnaire on paternal coping behavior
- LARES Geschwisterbogen

### Speech pediatric field:

- Parental questionnaire for early detection of at-risk children (Elfra 1)
- C-BILLT (Computer Based Instrument for Low Motor Language Testing)

### Music therapy:

- Diagnostics of communicative skills
- Initiation of speech

### PD Dr. Cornelia von Hagen

Dipl.-Psychologist

### Focus on the work of the scientific activity at the TUM:

- Development and evaluation of training programs for families with chronically ill children
- Participation in pharmacological studies

### Focus on work as project manager in the kbo-Kinderzentrum München Schwabing:

- Chronic physical illnesses
- Genetic Syndromes
- ADHD
- Language development disorders
- Neurofeedback

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