



Resuscitating a dream:

An anaesthesiologist is striving to improve access to rare disease information

Uta Emmig first considered the problem of accessing information for rare disease anaesthetics when she was a graduate student. Now a working anaesthesiologist, she is reviving her earlier pursuit of improving information for working with rare disease patients in her particular field.

Uta Emmig, a German anaesthesiologist who specialised at the University Hospital of Aachen and is now working in Italy, knows from firsthand experience that anaesthesiology information specific to rare disease patients can be hard to come by. She has spent many hours surfing the web for literature providing concrete information on administering anaesthetics or the necessity of specialised equipment specific to rare disease patients. Even when such information exists in the literature (via case reports, for example) if the hospital or institution does not have access to a particular journal, the information remains inaccessible.

It is one thing for an anaesthesiologist to know in advance that they will be working with a specific patient with a rare condition that will require special monitoring or equipment, but what about the emergency room professional who encounters a patient with a disease they know little or nothing about? To address this vital need, Dr. Emmig is working to improve the information available to professionals in her field. *OrphaNews Europe* met with Dr. Emmig recently and was able to learn more about this critical aspect of rare disease patient management:



OrphaNews Europe: How did you first become aware of the need for information for anaesthesiologists working with rare disease patients?

Dr. Uta Emmig: I have been studying the anaesthesiology information resources relevant to rare diseases since 1996. My thesis advisor (Pr. Manfred Abel, former professor of paediatric anaesthesia at the university hospital of Cologne, currently head of the anaesthesia department

in a teaching hospital in Cologne-Porz) first drew my attention to this problem. He actually created an electronic database for anaesthetics for paediatric rare diseases with the goal of offering a counselling service for professionals in the 1990s. Unfortunately, he had to abandon his efforts due to lack of funding and partners. For my thesis, I conducted a study of the literature as well as a survey amongst my colleagues. The outstanding result was that it was very difficult to find information.

OrphaNews Europe: What kind of information do anaesthesiologists need to work optimally with this category of patients?

Dr. Uta Emmig: For elective interventions/planned surgery:

1. Before the anaesthesia: “Do I have to perform other examinations?”
2. During the anaesthesia: “Do I have to monitor/control organ functions that I usually wouldn’t control?”
3. After the anaesthesia: “Do I have to keep a watch on the patient for a longer period than usual? Do I have to monitor other organ systems, e.g. blood glucose level.

For emergency surgery:

1. “Can I get rapid access to information on a specific rare disease?”
2. “How can I know which possible medications a patient might be taking and access detailed information concerning interactions with anaesthetic agents?”
3. “Does the hospital or clinic where I am working have specialised equipment available if necessary? If not, where can I access such equipment?”

OrphaNews Europe: Can you comment further on emergency care anaesthesia in relation to rare disease patients? What are some examples of particular information that might be needed in this type of scenario?

Dr. Uta Emmig: Particular information involves hidden problems that the health professional cannot see but which can result in severe complications, such as a metabolic disturbance or instability of the upper respiratory system.

OrphaNews Europe: What kind of information exists to date? Is there any centralised resource that gathers information on anaesthesia for rare disease patients?

Dr. Uta Emmig: There are a few resources that present an overview of the subject. Foremost, there are two brilliant textbooks:

Anesthesia and Uncommon Diseases by Lee A. Fleisher (WB Saunders, 2005, ISBN 1416022120)

Anesthesia & Uncommon Diseases by Jonathan Benumof, ed. (WB Saunders, 1998, ISBN 0721686737)

Another work on the subject is: *Anesthesia and Co-Existing Disease* by Robert K. Stoelting and Stephen F. Dierdorf (Churchill Livingstone; 2002, ISBN 0443066043)

I also had the possibility to contribute my thesis work to a textbook:

Anaesthesie bei seltenen Erkrankungen (Anaesthesia in rare diseases) by Peter Biro, Dierk Vagts, Uta Schultz and Thomas Pasch (Schultz is Dr. Emmig's former name) (Springer Medizin Verlag, 2004, ISBN 3-540-00634-6)

In addition, there is information available via Medline, NORD, (the US-based National Organization of Rare Diseases), and Orphanet. But to my knowledge, none of these resources currently has a database specific to anaesthesia and rare diseases. Perhaps there is such a resource in Japan. My literature research in 1996 revealed the existence of many case reports in the *Japanese Journal of Anaesthesia* (Springer Japan).

There are also various projects, available via internet, which have initiated a anaesthesia and rare diseases subunit, but most of these projects (usually the work of an individual or single institute, rather than a collaborative effort) are unfinished and eventually abandoned.

OrphaNews Europe: Could you describe your idea for the OrphanAnaesthesia project?

Dr. Uta Emmig: OrphanAnaesthesia is an information-based project proposal that would give anaesthesiologists free access to relevant, peer reviewed information to ensure the quality of anaesthesia delivery for rare disease patients. It would also include a posting site on which anaesthesiologist could report their experiences with anaesthesia for rare disorders. The overriding goal of the project would be to increase the amount of available information.

OrphaNews Europe: How could information on anaesthesia for rare diseases best be gathered and made available?

Dr. Uta Emmig: We have proposed collaboration with Orphanet because we found it has an excellent existing database. To begin with, this collaboration might take the form of creating an *Orphanet Series Report* document that could address the issue of anaesthesia and rare diseases. Orphanet is also currently producing emergency care guidelines for specific diseases. Perhaps we could contribute our expertise to this effort.

OrphaNews Europe: How would you envision organising data in view of the large number of rare diseases that exist?

Dr. Uta Emmig: As I mentioned above, I think a collaborative effort with Orphanet, using the existence of the Orphanet database, would afford the best organisational method for storing anaesthesia information specific to rare diseases.

OrphaNews Europe: What kind of collaboration or support do you currently have from amongst your colleagues?

Dr. Uta Emmig: The Paediatric Anaesthesia Scientific Working Group (Wissenschaftlicher Arbeitskreis Kinderanästhesie or WAKKA) of the German Society of Anaesthesia and Intensive Care – (Deutsche Gesellschaft für Anästhesiologie und Intensivmedizin or DGAI) discussed the possibility of creating a database with rare disease information some time ago. In May 2007, I presented the idea of collaborating with Orphanet. My ideas were well received. In particular, the current chair of the group, Pr. Dr. Jochen Strauss (Department of

Anaesthesiology and Intensive Care - Helios Klinikum Berlin) was supportive of the project. The DGAI president, Pr. Dr. Hugo van Aken of (Department of Anaesthesiology and Critical Care, Universitätsklinikum, Muenster) considers this project as visionary for Europe.

I personally hope fervently that all the European societies of anaesthesia will join this project. The OrphanAnaesthesia project proposal was presented in Amsterdam in September 2007 at the annual meeting of FEAPA (Federation of the European Associations of Paediatric Anaesthesia.). We received much positive feedback on our ideas, such as the Finnish professional in the field, who exclaimed, "Finally!" – expressing the sentiment that he and others had been waiting for a source of information such as this.

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