Our Child Is Born … Disabled

Joy and sorrow sometimes go hand in hand. Nobody knows that better than the parents of a disabled child. At first, when it becomes apparent that there might be something wrong with a newborn baby, they experience a growing sense of fear. Then comes the feeling of despair when it emerges that there really is something wrong. A clear diagnosis can bring enormous relief because the problem is given a name and an identity. However, when the parents begin to understand the diagnosis, fear takes over again. They start to imagine what it might mean for their future lives, their marriage and the development of the whole family. Finally comes the inevitable question 'Why us?'. Only then does the family begin to look forward and tackle the future.

With Prader-Willi syndrome (PWS), joy and sorrow continue to live side by side because the development of children with PWS is affected at all three levels: biological, psychological and social. Good times are frequently followed by difficult ones, with all the family's resources often stretched to the limit.

The aim of this booklet is to make life a little easier for parents, relatives, doctors and therapists of children with PWS by giving them a brief, comprehensible overview of what is currently known about this condition. The Swiss PWS Association first asked me to put something like this together a few years

‘Of course it is important to know what PWS is and what the related problems are. But reports often forget to mention the “positive” side. Who of our friends, for example, could so easily take their baby to a restaurant without it constantly crying, as we can with our quiet Pierre? Most importantly, it is vital always to remember the joys you can experience with your child!’

ago. An initial edition was produced in 1998, based mainly on the experiences of parents from the Swiss PWS Association. In the last 5 years, however, research into appetite regulation in general as well as therapy for children with PWS have developed so much that it became necessary to revise the booklet completely and to make it internationally accessible by having it

translated. I should point out that the latest scientific findings are also described in two comprehensive books which are currently available. Interested readers will find that they contain detailed information about topics which are only covered in general terms here. Despite – or perhaps because of – the tremendous progress in research on PWS, this booklet should be treated as a project which is permanently under construction rather than a finished product.

This publication is divided into six sections. After the introduction, the second chapter provides an overview of the history of research into the syndrome, its main characteristics and the most important methods of treatment. The third part describes the genetic causes, explains how the genetic defect is translated into the symptoms, and discusses the diagnostic process. The fourth and fifth sections contain detailed descriptions of the symptoms and methods of treatment, which are followed by a brief conclusion. In order to illustrate the everyday life of parents of children with PWS, we have added some excerpts from conversations with the parents of some of my patients.

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