Rubinstein-Taybi Syndrome: A Book for Families

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Introduction

This book is designed to answer some of the questions which parents frequently ask about Rubinstein-Taybi syndrome. Since this is a relatively rare condition, there are still many unanswered questions. It is important to realize that each child with Rubinstein-Taybi syndrome is unique and that abilities and handicaps are variable. Additionally, not all medical problems which are described in this booklet apply to every child.

"Travis is an absolute joy in our lives. When he was born, the devastation that I felt kept me from understanding what a wonderful part he and yes, even his small delays and handicaps would be in our lives. He is very loved by us all -- including everyone at his school and everyone at our church. I still have moments when I wish he were "normal" and could have more of a "normal" life. I am sure that this will never completely subside but I have come to love and appreciate Travis for what he is instead of dwelling on what he isn't. With the acceptance of these problems comes such an overwhelming appreciation and desire for him to simply reach his potential. He has become an important part of our family. He is happy and so are we."

What is the Rubinstein-Taybi syndrome?

Rubinstein-Taybi syndrome refers to a specific pattern of physical features and developmental disabilities which occur together in a consistent fashion. Individuals with Rubinstein-Taybi syndrome have short stature, developmental delay, similar facial features, and broad thumbs and first toes. The condition was first described in 1963 by Dr. Jack Rubinstein and Dr. Hooting Taybi who recognized this pattern in seven unrelated children. Since 1963, there have been over 400 individuals reported with the Rubinstein-Taybi syndrome. Although the exact incidence is unknown, it has been estimated that approximately 1 out of 300,000 persons has the Rubinstein-Taybi syndrome. The condition occurs with equal frequency in males and females.

How do we know someone has Rubinstein-Taybi syndrome?

Most children with Rubinstein-Taybi syndrome have a physical appearance which is different from that of their family background at the time of birth. The average age at the time of diagnosis of Rubinstein-Taybi syndrome is approximately 15 months. It is often difficult to be certain of the diagnosis at birth, and many children are not recognized for several months or even years. There are no medical tests which will aid in specifically diagnosing the Rubinstein-Taybi syndrome. The only way to know if someone has the condition is by noting the characteristic physical features. The facial features include a small head size, thick scalp hair which may extend onto the forehead, down-slanting eyes, prominent nose, small mouth, and a high-arched palate. Additionally, the thumbs and first toes are broad and sometimes angulated. Many of these physical characteristics have little significance other than as clues for the

diagnosis. The features may not be obvious to family members or friends, but should allow the diagnosis of Rubinstein-Taybi syndrome to be made by a specialist in clinical genetics and often by a pediatrician.

"Of all I went through, the area that I feel most strongly about is the manner in which I was told about Scotty's diagnosis. This occurred when Scotty was 12 days old. The doctors asked if they could take Scotty into another room to examine him. We were not allowed in the room. In five minutes they came back and handed my mom and me a yellow Kleenex (I still won't buy yellow Kleenex) and the doctor began telling us that Scotty had a rare genetic disorder called R_____. That is all I could understand because she was talking so fast and she rattled on and on about how he would never read and he would be trainable mentally retarded and that some parents have chosen institutions for children like these. I feel that I was given too much information for any human being to handle emotionally. I was only given the worst news and not a sign of hope for anything good. We have come such a long way since then. Yes, Scotty has a low IQ but he is not trainable, he is educable. Yes, Scotty is non-verbal but he understands signs and I think he will talk. We have no plans now or anytime for institutionalizing him. He is such a joy--a little bundle of giggles, love and simplicity. When he is 18, we will help him to decide what he wants to do. That is too far off to worry about now. We haven't taught Scotty half of what he has taught us. He loves laughs and enjoys so intensely. We are blessed to have him--for however long God chooses."

What causes Rubinstein-Taybi syndrome?

The cause of Rubinstein-Taybi syndrome is unknown. A chromosome abnormality has not been identified in this condition. However, it is possible that there is a spontaneous change (mutation) in a gene which is too small to be detected by chromosome analysis. There have been no consistent chemical or other environmental exposures reported during pregnancy. There is no reason to believe that anything the parents did or did not do during the pregnancy caused the child to have Rubinstein-Taybi syndrome. Additionally, no definite genetic pattern has been identified.

"Our son, Steve, was diagnosed as having Rubinstein-Taybi syndrome within hours of his birth. He was born in a large medical center which facilitated the diagnosis but also meant that he was the source of great interest to every teacher, student, therapist, geneticist, etc. within reach. We were shocked, hurt, disappointed, and angry--everything but happy at the birth of our first child. Thankfully, the dire predictions were off base. Steven is just over 3 years now and in a preschool handicapped program five mornings a week. He is a happy, delightful, attractive child whose most serious health problems are chronic congestion and a stubbornly infected ingrown toenail. Life with Steven is not sorrowful. At times, it is bitter-sweet and for the most part, our lives are as"normal" as any modern family. We just enjoy what we have together and hope life continues to be kind to us and to Steven."

Is Rubinstein-Taybi syndrome inherited?

In almost all cases, there are no other family members with the Rubinstein-Taybi syndrome. There have been seven reports of familial occurrence in the medical literature, however, most are poorly documented or the descriptions have been incomplete. Of these, two are fairly well documented familial cases, one in which a brother and sister both have Rubinstein-Taybi syndrome, and one in which a mother and son both have the condition. Based on the small number of recurrences in families, the risk to have a second child with Rubinstein-Taybi syndrome is probably less than 1%. However, the risk for a person with Rubinstein-Taybi syndrome to have an affected child is probably much higher than, possibly as high as 50%. At the present time, there is no way to test a baby prenatal (before birth) to see if he/she has the condition.

What can we expect in the newborn period?

Most babies with Rubinstein-Taybi syndrome are born at term after an uncomplicated pregnancy. However, over 1/3 of pregnancies are accompanied by polyhydramnios (excessive amniotic fluid). About 1 out of 5 babies with Rubinstein-Taybi syndrome will require treatment in a newborn intensive care unit. Medical problems during this time may include breathing difficulties, feeding problems, poor weight gain, and severe constipation. A neonatologist (specialist who takes care of newborns) usually manages the care of the infant and keeps the family informed of his/her progress.

"Eight years ago when our daughter was born, her future was very scary to us. So many things we didn't know and so many things we knew we had to face. Times have been very hard and trying, but our little girl has brought us tremendous joy. Some people label her different and developmentally delayed, we also see her as outgoing, full of love, and a willing friend to anyone. We don't know what her future will be but we plan to do everything possible to her advantage and help her to live a 'normal' life.

What medical problems can we anticipate?

Much of the medical information presented in this book was obtained through a questionnaire of 50 children and young adults who belong to the American Rubinstein-Taybi Syndrome Parent Group. The study group consisted of 29 females and 21 males who ranged in age from 1 to 26 years. All of these individuals were being reared at home. For this reason, we feel that our data regarding the medical problems, intellectual capabilities, and behavioral characteristics of persons with Rubinstein-Taybi syndrome are more applicable to families than previous information obtained from institutionalized individuals.

A variety of medical problems can be seen in children with Rubinstein-Taybi syndrome. However, it is unlikely that an individual child would have all of these difficulties. The majority of children experience poor weight gain in early life primarily due to feeding

problems including vomiting and swallowing difficulties. In some cases, this is severe enough to require hospitalization for failure to thrive. However, these feeding problems tend to resolve during the first year of life.

"Jeremy had feeding problems which included the inability to coordinate sucking and swallowing. He would spit out as much as he would suck. He needed to be gavages (tube) fed as long as he was in the hospital. We took him home and fed him small quantities frequently and he gained weight, but very slowly. He has always been small, but in the last year he has grown 2 inches and is now very sturdy and husky."

Over 80% of children with Rubinstein-Taybi syndrome have some type of eye abnormality including strabismus (crossed eyes), cataracts, and tear duct obstruction. Glaucoma may be present at birth or in early life which can result in blindness if undetected. Therefore, it would be advisable for every child to have an eye examination by an opthalmologist.

Frequent ear infections are seen in about half of patients with mild degrees of hearing loss being seen in 1/4 of patients. Recurrent upper respiratory infections are also common in infancy and childhood.

Dental problems are present in 2/3 of children, most commonly due to overcrowding of the teeth and an increased frequency of cavities. A high-arched palate is also present in most individuals with Rubinstein-Taybi syndrome. Orthodontic treatment is necessary in most patients. A high percentage of individuals have talon cusps on the underside of the permanent incisors. A talon cusp is an accessory cusp-like structure which may contain pulp. In some patients they may result in cavities or irritation of the tongue requiring treatment, while in others no problems are noted.

Congenital heart defects are present in 35-40% of people with Rubinstein-Taybi syndrome. Some of these defects are mild while others are more serious and may require surgery.

Most boys with Rubinstein-Taybi syndrome have undescended testicles, and almost always surgery is required to correct this. Severe constipation can be a very difficult problem in Rubinstein-Taybi syndrome. Many children require dietary modifications, enemas, and suppositories, but it is best to discuss this with the child's physician.

"Kurt suffered so much with the constipation problem from 9 months of age to 2 years of age. Lower GI tests were done along with biopsies, but nothing was found except thick muscle in the wall of the intestine. After using about everything under the sun from milk of magnesia, suppositories, enemas, fiber additives, and physically having to help him pass a bowel movement, we found that daily yogurt is what helps Kurt."

Broad thumbs and first toes are seen in virtually all patients with Rubinstein-Taybi syndrome. The fingers may also be somewhat broad. When the thumbs are also angulated, surgery may be required to improve their use. A referral to a hand surgeon

experienced in this type of surgery should be made. Very broad first toes may require surgery to increase comfort while wearing shoes. Other potential orthopedic problems include dislocated patellas (kneecaps), and scoliosis (curvature of the spine). A physician can tell by examining the child whether or not these problems are present.

There are several reports of keloids (excessive scarring) in individuals with Rubinstein-Taybi syndrome. These large scars may form after surgery, trauma, or chicken pox.

There may be some increased risk associated with anesthesia (particularly involving arrhythmias) in Rubinstein-Taybi syndrome. Although this issue warrants further investigation, parents and anesthesiologists should be aware of these potential problems.

We have not described every potential medical problem which may occur in Rubinstein-Taybi syndrome. We also want to emphasize that most children will not have every problem which has been discussed. Some children have chronic medical problems while others have minimal difficulties.

Should my child be immunized?

In our study group, over 400 immunizations were given. The overall complication rate was only 5%, which is no greater than the general population rate. Reactions consisted primarily of low grade fever and irritability. Thus, there seems to be no reason to withhold immunizations in these children. As in all children, immunizations are important in order to prevent serious infectious diseases.

Growth

Most children with Rubinstein-Taybi syndrome are of average weight and length at birth. However, within the first few months, growth falls well below average. Children with Rubinstein-Taybi syndrome are short throughout life and do not seem to have the usual growth spurt around puberty. The average adult height in males is 5', while in females it is 4'10". However, the ultimate adult height is variable and is probably also affected by family background. Boys tend to be overweight during school-age years, while girls may be overweight in adolescence.

Puberty and Fertility

In our study, the onset of puberty occurred at approximately 12 years of age, which is similar to the general population. Young women began to have menstrual cycles at the usual age as well. We are aware of two women with Rubinstein-Taybi syndrome who have been reported in the medical literature to have had children. One woman had a son affected with Rubinstein-Taybi syndrome and a healthy daughter. The other woman had an unaffected child. We must assume that individuals with Rubinstein-Taybi syndrome are physically capable of having children.

Development

All individuals with Rubinstein-Taybi syndrome have developmental delay; however the degree of limitation is variable. In our study of home-reared children, the average IQ (intelligence quotient) was 51, but ranged from 30-79 (normal range is 85-115). One boy in our study had a non-verbal IQ of 82. In general, non-verbal test scores were higher than those which required good verbal skills. Thus, the average person with Rubinstein-Taybi syndrome functions in what is referred to as the moderately retarded range. Some individuals may be more severely retarded while others fall into a borderline normal category. The highest reported overall IQ in a child with Rubinstein-Taybi syndrome has been 80. Previous reports in the medical literature indicate lower IQ scores than in our study, however most of these studies involved institutionalized patients. We feel that our study group is more representative of home-reared handicapped children and is more representative of home-reared handicapped children and is of more value in predicting the potential of children with the Rubinstein-Taybi syndrome. However, it is difficult to predict the abilities of an individual child since each certainly has his/her own strength and weaknesses.

"Angela is considered mildly to moderately retarded so she is in a self-contained classroom with 4 other children, a full-time teacher, and an aid. She goes with her non-handicapped peers for special activities such as art, music, home economics, shop, gym, and homeroom. She gets along great and has lots of friends. Last year she even helped with the props for the class play and was a basketball manager for the girls' intramurals."

Speech problems are present in about 90% of patients. Most individuals have speech delay, but others also have articulation problems. Six percent of our study group used sign language exclusively for communication while 50% used sign language in combination with speech. Most children require formal speech therapy.

All children with Rubinstein-Taybi syndrome continue to make progress and learn new skills throughout life. Developmental milestones are indicators of a child's progress and learning. The table below lists the average age and the range of ages for the attainment of common motor and language milestones by the children in our study.

Skill Average (months) Range (months) Normal Range (months) rolled over 7 2-24 2-5 crawled 15 8-30 7-10 sat up 11 6-30 5-8 walked 30 15-54 11-15 first words 25 6-57 9-13 3-word phrases 65 24-156 14-24 toilet trained 63 30-216 24-27 rode tricycle 68 42-216 36-48 Most children with Rubinstein-Taybi syndrome begin therapy or preschool at a very early age. The majority of these children receive both speech and physical therapy in addition to special education. Several options are available including early intervention programs and other preschool programs for handicapped children. These options and facilities will vary with each individual community. Older adolescents may receive vocational training in preparation for sheltered or supported employment. Some adults with Rubinstein-Taybi syndrome are able to live semi-independently while others may remain with their families.

"Our daughter's progress has been achieved through private therapy sessions, speech, occupational therapy, physical therapy, and music therapy. We made the

startling discovery that she can learn through music more easily than any other method. Hence, music therapy has been our lifeline for education and also for working on behavior and social skills."

Personality

Each child with Rubinstein-Taybi syndrome is a unique individual with his/her own personality. However, in general, children with Rubinstein-Taybi syndrome are described by their families as loving, friendly, and happy as opposed to irritable or unpleasant. Special interests of the children in our study included music, swimming, looking at books, and watching television.

"Sheri has been involved in swimming programs, Saturday Recreational Program, Pegasus (therapeutic riding program), Special Olympics, and special ed church school. Last summer she won the gold medal in the equestrian events. Sheri has a job three days a week during school hours. She works at a travel agency doing a variety of tasks."

Behavior

Sometimes behavior in a handicapped child can be the greatest concern of his/her parents. Some children with Rubinstein-Taybi syndrome engage in occasional self-stimulatory behavior such as rocking, spinning, and hand-flapping. A short attention span was described in 90% of our patients, but only a few have been treated for hyperactivity. About half of our patients were said to react differently to sound. Many of the children seem to dislike loud sounds or do not tolerate crowds because of noise. The child's doctor or teacher may be able to recommend a behavioral specialist or psychologist who can make suggestions about coping with behavioral problems. In some instances, an individualized therapy program or trial on medication may improve difficulties with maladaptive behavior.

"Kurt is a happy, energetic little boy with a big heart and a strong spirit. He thrives on attention and his smile makes everyone feel good. Kurt loves to hug, touch, feel, manipulates--he needs lots of hugs, stroking, and reinforcement. He always has a smile for everyone and has touched the hearts of many. We have used time-out for negative behavior or we attempt to move him through proper behavior. Kurt's ability to tell us what he wants with signs has helped tremendously and has reduced his frustration level a great deal. He understands and can do a lot more than people think he can."

Organizations which may provide information:
March of Dimes Birth Defects Foundation
1275 Mamaroneck Avenue
White Plains, NY 10605
(914) 997-4624

The Alliance of Genetic Support Groups

38th and R Streets, NW
Washington, DC 20057
1-800-336-GENE

National Organization for Rare Diseases (NORD)
P.O. Box 8923
New Fairfield, CT 06812
(203) 746-6518

Association for Retarded Citizens (ARC)
2501 Avenue J, P.O. Box 6109
Arlington, TX 76005
1-800-433-5255

"We had the opportunity to meet another child with Rubinstein-Taybi syndrome and it was amazing to see the similarities, right down to facial expression and favorite things like music."

"It has helped us tremendously to get to know other parents with children who have Rubinstein-Taybi syndrome and to hear from someone who is dealing with the same problems, etc. We also realize through this contact that all the children with Rubinstein-Taybi syndrome are individuals and even though they do share a lot in common, they also have differences. We can always search and look for answers to our questions and in years to come, there will be answers. For now, we will work on what is happening here, trying to get through each day, one at a time. We will always want more for our child and us, but we can also see what has been given to us and the blessings that we have received."

Additional copies available:
Rubinstein-Taybi Book
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http://www.rubinstein-taybi.org/ -- Revised: January 15, 1997