



Rubinstein-Taybi Syndrome

An Information Booklet

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Note: This booklet was written in the UK for UK-based families, and practices and provision of facilities may be different in other countries.

The contents of this booklet have been written by parents and carers from the Rubinstein-Taybi Syndrome Support Group, using their own experience as well as information provided by acknowledged medical and psychology professionals. They have aimed to ensure, to the best of their ability, that any facts given are as accurate as they can be given the current state of medical knowledge about the syndrome. All content is provided in good faith, and the Support Group cannot accept responsibility for any errors, inaccuracies or omissions, or for the results of anyone acting or not acting upon any statement in this guide

Foreword

This booklet has been produced by the Rubinstein-Taybi Syndrome Support Group, a UK registered charity. Formed in 1986, the group offers information and support to families and individuals affected by Rubinstein-Taybi Syndrome (RTS).

Membership is open to individuals with RTS and their parents/carers, and is free of charge. Regular get-togethers and family weekends feature among the group's activities, and provide families an opportunity to share experiences and friendship. Our twice yearly newsletter ensures that families are kept up to date with current information, events, information and news from other families etc. All of this is financed by voluntary donations and fund-raising. If you would like to find out more, make a donation, or become a member, please visit www.rtsuk.org.uk/contact/.

Much of the information available about Rubinstein-Taybi Syndrome (hereafter RTS) is in the form of papers and articles written by medical professionals, and as such is impersonal, and uses medical terms and language that are not readily understood by the lay person. The management committee of the RTS Support Group realised there was a need for information for families and carers which was easily-readable, and which included the personal experiences of people living with the syndrome on a daily basis; so we created this booklet. It will be of particular use to parents/carers whose baby or child has been newly-diagnosed but also, we hope, to those with older children, young people and adults with the condition. In addition, we know that many people find it useful to provide a copy to each professional involved in their child's care, for example, the paediatrician, support worker or teacher.



Henry and his mum, Jo

Henry was very poorly from birth with lots of different problems. Initially we were in shock and completely devastated. When his diagnosis was confirmed we didn't want to believe it and couldn't face meeting anyone with the same condition. Henry was just our baby not a syndrome.

Information about the support group was the only genetic counselling we got. The booklet was brilliant and the website is always available. Over the years I've started using the Forum, and finally attended a get together this year. Henry's now six and the RTS Support Group have been a constant companion in our lives and will continue to be so. Even in the darkest of times, access to the group has meant we've never been alone. The RTS Support Group is our extended family, better really because we choose to have them in our lives! Like the best of friends, the group never imposes and is always ready, willing and able to help. Simply there...whenever you need them. Experience breeds the best advice; the support group has families at all stages of our special journey with our extraordinary children.

No one has the perfect map... but the RTS group are a fantastic route finder!

Henry's mum

Introduction

What is Rubinstein-Taybi Syndrome?

RTS is a specific pattern of physical features and development disabilities that occur together in a consistent fashion.

A syndrome is a group of features that together characterise a medical disorder.

In 1963, Dr Jack Rubinstein (a paediatrician) and Dr Hooshang Taybi (a radiologist) examined seven children with an intellectual disability, short stature, broad thumbs and broad big toes. Following diagnosis, the syndrome was initially named the 'broad thumb-hallux syndrome'. Later researchers re-named the syndrome after the two founding doctors, hence, Rubinstein-Taybi Syndrome.

It has been estimated that the syndrome occurs in one of 100,000 to 125,000 babies, depending on the population being studied. It occurs in all races and equally between boys and girls. Since the syndrome has become readily identifiable there are more than a thousand cases reported from around the world.



Why does it happen?

At present it is not possible to detect RTS in routine screening during pregnancy.

An abnormality of chromosome 16 or chromosome 22, meaning they are damaged, missing or untraceable, is thought to be the cause of this collection of features and disabilities called RTS. There is no evidence to suggest that anything done or not done, before or during pregnancy causes a child to be born with RTS and there is no known cure.

Is RTS hereditary?

RTS is present at birth and is caused by a change in a genetic trait. It is usually not inherited from one of the parents but started in the child. The possibility of producing a second child with the syndrome in the same family is only minimally raised and the chance of this happening is certainly much less than 1%.

Multiple births may result in all the babies having RTS if they are identical, but apart from these instances, there is only one family that is known to have two children with the syndrome. The chance of siblings going on to have a child with RTS is no greater than that for the rest of the population.



Sarah, Ava and Freya

If someone with RTS develops to such a level that he or she is considering having children, then the chance a child is again affected is about 50%. If the cause of RTS in the parent is known before the pregnancy, it would be possible to check for it by pre-natal diagnosis, but if the cause is unknown no reliable prenatal diagnosis is possible.

After diagnosis it is extremely difficult to tell parents how their child will develop. What is clear is that a baby with RTS will develop into a toddler, schoolchild and adult, the great majority of children will laugh, talk, crawl, walk, ie have a degree of physical activity, but there will be delayed development.

RTS children love contact especially with their parents, siblings and other relations. They love to love and be loved; they tend to be happy and fun loving but sometimes sad, angry and stubborn; the majority will learn to eat, to wash and go to the toilet independently; they may be able to dress and undress themselves with or without help. Many learn to cycle (some with special adaptations), swim, and a love of horse riding (supervised and assisted) is often a great source of pleasure. Some will learn to read and write, albeit at a lower level. They enjoy hobbies and friends; they may join youth groups, like Brownies or Cubs who can accommodate special needs. A love of music is common in many children with

RTS. It can provide great enjoyment and be of therapeutic value at times, even if learning to play an instrument proves impossible.

It has often been said, “a child with RTS can bring out the best in other people.” They enjoy life, and have a magic ability to make you smile, even if you don’t feel like smiling! They need to be given as much opportunity as possible to increase their contact with the world outside their own home and family, and to experience “normal” interests and activities. The more experiences a child has, the greater his or her chance to reveal hidden talents will be.



Henry and Daisy

Diagnosis



Boyd

“Parents are people who have children; parents of children with special needs are no different from other parents. They come in all shapes and sizes, ages, walks of life, backgrounds and cultures ... They are simply parents.”

Often there are no serious problems and specialist referral occurs when the parents notice delayed developmental milestones eg; sitting, crawling, talking. Sometimes it is possible to confirm diagnosis through specific chromosome investigations but usually observing the child and listening to the parents' story makes it.

Most children are born full term, usually after an uncomplicated pregnancy. The birth weight and height tends to be around average. Some mothers have reported that they have had high blood pressure, excessive amniotic fluid and increased foetal movement or decreased foetal movement.

The babies are often quiet, passive and tend to sleep a lot. The baby usually has a round face, a small head size, abundant dark hair, a prominent nose, small mouth and a high palate. Sometimes they are born with a temporary red birthmark on their forehead; nearly all have broad and sometimes angulated thumbs and big toes.

Some parents are given the news that their child has a disability at birth. Others receive a diagnosis after months or years, often knowing or suspecting that 'something was wrong' from an early age. You may be given, or find, a lot of negative information about RTS, such as a number of things that their child "won't be able to do", or a list of medical conditions associated with the syndrome. If that is

the case, it can help to remember that seldom do people with RTS have all the conditions commonly listed (some have very few); and no-one can predict with any certainty what limitations to their abilities your child will eventually have.

Depending on the timing of the diagnosis and the way in which you discovered it, your feelings and reactions will vary in nature and intensity. Parents go through many of the same emotions that would be felt had they experienced bereavement. This includes feelings of grief, shock, disbelief, denial, anger, isolation and then acceptance.

If you were told at, or shortly after birth that your baby had RTS, the shock and grief may be particularly intense. This is because when most parents are expecting a baby, they are expecting a 'normal' and 'perfect' baby. After nine months, adjusting to a baby whether or not he/she has RTS, is a major task. If you have been told that your baby will have extra needs, this period of adjustment will be even greater.

If you were told sometime after the birth, you may feel some relief that eventually someone is listening to you, that you are no longer labelled as being a neurotic parent, and that you have a name to put to your child's condition.

Parents, like their children, are all different. Some want to know the minimum and others can't get enough information. It is important that you ask questions when you feel you are ready, as you may not want to do this straightaway. Remember that being told your child has RTS will probably have come as a shock and may be difficult to deal with. At this time your capacity for taking in all that is said is very limited.

“Our daughter was not diagnosed as having RTS until she was around 5 and a half years old, as the more obvious signs of the syndrome only became apparent as she got older.

The first inkling we had that there might be “something wrong” was when she was referred to a paediatrician at the age of 8 months, due to the small circumference of her head. He simply decided to monitor her growth, but after that, it gradually became apparent that she was reaching her milestones later than average as well. So initially it was the professionals - health visitor, paediatrician, etc - who realised that she might have some kind of special needs, while Dad and I were in denial, looking for reasons why she might not have developed at the average rate, such as having frequent chest infections. Chromosome tests came back as normal and it was only as her facial features developed, that the diagnosis of RTS was made. By that time we knew our daughter had a learning disability, and felt it was a positive step finally having a name to put to her condition.”



Katie

Katie's Mum



Steven

“When Steven was born he had some problems with his breathing and needed to go to the special care baby unit.

During this time the doctors suspected that he had RTS but did not say anything to us at that time.

We took him home when he was 3½ weeks old not aware that he had RTS and would have learning difficulties. When he was 8 weeks old I took him back to the hospital to see the paediatrician and he told me that he thought that “he had RTS and would be a slow learner”. I was in a state of shock and didn’t fully realise the implications, all I knew was that I loved Steven more than anything and what ever having a son with RTS meant, he was my baby and I would find out as much as possible about the syndrome and where I could get help. His diagnosis was confirmed by the consultant geneticist when Steven was 5 months old. I feel that even though the doctors knew that he had RTS at birth, in hindsight I was glad I was not told until I had taken Steven home from the hospital and had got over his initial difficulties.”

Steven’s Mum

“Our son John was approximately three when we received the diagnosis. John had an operation to correct a squint in both eyes. We were told at the time it is a very common operation and many children go through the procedure. Little did we know that the following visit for a check up was to bring us the most shattering news that we had received since John’s birth.

Even though we were told that when he was born there was “something wrong”, we never received a diagnosis. So it came right out of the blue and was a tremendous shock to us both. I remember John after the operation playing in his play pen in the hospital with patches on his eyes. He did look so vulnerable.

Sometime afterwards we had a follow up appointment at the hospital to check his progress. We were asked if some students could sit in on the consultation. We agreed and sat down in front of the consultant and his colleagues.



John

John sat on my knee. What we were about to hear came as a shock to both of us. The consultant started to speak to the students and outline the operation that John had endured.

He continued with, “John is a typical Rubinstein”.... That was all I heard. The rest of the description of John’s condition I didn’t hear. I stopped him immediately, “Could you please tell us the diagnosis before explaining it to your colleagues?” I remarked. The consultant looked surprised. “You mean you don’t know what your son has been diagnosed with?” he replied.

I informed him that no one had told us about any syndrome relating to John's condition and that all we knew is on the day of John's birth the nurse had remarked that our son John "was not right". We left soon after the conversation. We were both in a daze. It was another black hole and it was the beginning of another chapter in our lives.

Looking back when we were outside the hospital Kath asked me, "What was the name of the syndrome that the consultant spoke about"? I tried to make out that I had forgotten the name. Kath would have none of it. "What was the name of the syndrome John?" she repeated. "I only caught the first name, Rubinstein". The rest got lost in the shock of the announcement. Here we go again, in a feeling of bewilderment and isolation. Where do we go from here?

A few years later we would contact the support group which enabled us to gain knowledge and share experiences of the syndrome and make some friends who were dealing with the same problems. This was to be one of the most positive moves that we have ever made. The

support and understanding we receive is "priceless". Kath and I have made many friends with people all over the UK.

The support group work tirelessly to support families and carers, most of them are parents of children with RTS so they know what families are going through".

John's Mum and Dad



John

Reactions and Emotions

Reactions and emotions experienced by all or some parents, and at different times include:

Denial, shock, numbness, reflection

This is an immediate reaction to the news. It will take time to mentally adjust and understand the new circumstances while initially not wanting to believe that your child has RTS.

Euphoria, minimising the problem

This is an early stage of acceptance; feeling positive about the situation, seeing hope and making the best of the situation. feeling positive about the situation, seeing hope and making the best of the situation.

Pining, searching

For a period the parent(s) may go back, in reality of thought – was there something we could have done? Should we have known something was wrong?

Anger

Not having control or a choice over the situation can often lead to anger and frustration. There may be feelings of wanting to blame someone.

Guilt

Many parents experience a great sense of guilt about having a child with RTS. Parents should not feel guilty for there are no facts whatsoever to support the idea that you could have prevented your baby being born with RTS. It is a genetic disorder. There may be self blame, or your partner may think that you did something, or ate something when you were pregnant. Other siblings and

grandparents may also experience the feeling of guilt.

Depression, apathy

For a time you may feel that you have no motivation, getting up in the morning is an effort, interest in the baby/child is less. This can express itself through feelings of pessimism and can lead to difficulties in eating and sleeping. You may find yourself crying and losing interest in life and the people around you. If you think you are suffering from depression, it is important to seek professional help eg your GP.

Grief

Feeling sadness and grief is totally understandable and must be expected. These feelings are very similar to those experienced when you have lost someone close to you. Although you have not lost a child in terms of his or her life, you have lost the child you hoped for and you have to change the expectations you have for him/her.

Acceptance

Letting go and taking up the challenge. It takes time to come to terms with all these feelings but gradually it does happen. It is important to talk about how you feel with someone who understands. A listening ear can be found within the RTS Support Group, but there is sometimes a need for more professional help – perhaps the family doctor.

Many of the reactions and emotions seem negative but remember with time and support, the vast majority of parents do cope. Children with RTS – like all children – are both challenging and rewarding: it is an experience that puts life in perspective.

Welcome to Holland

by Emily Perl Kingsley

I am often asked to describe the experience of raising a child with a disability - to try to help people who have not shared that unique experience to understand it, to imagine how it would feel. It's like this.....

When you're going to have a baby, it's like planning a fabulous vacation trip - to Italy. You buy a bunch of guide books and make your wonderful plans. The Coliseum. The Michelangelo David. The Gondolas in Venice. You may learn some handy phrases in Italian. It's all very exciting.

After months of eager anticipation, the day finally arrives. You pack your bags and off you go. Several hours later, the plane lands. The stewardess comes in and says, "Welcome to Holland."

"Holland?!?" you say. "What do you mean Holland?? I signed up for Italy! I'm supposed to be in Italy. All my life I've dreamed of going to Italy."

But there's been a change in the flight plan. They've landed in Holland and there you must stay.

The important thing is that they haven't taken you to a horrible, disgusting, filthy place, full of pestilence, famine and disease. It's just a different place.

So you must go out and buy new guide books. And you must learn a whole new language. And you will meet a whole new group of people you would never have met.

It's just a different place. It's slower-paced than Italy, less flashy than Italy. But after you've been there for a while and you catch your breath, you look around.... and you begin to notice that Holland has windmills....and Holland has tulips. Holland even has Rembrandts.

But everyone you know is busy coming and going from Italy... and they're all bragging about what a wonderful time they had there. And for the rest of your life, you will say "Yes, that's where I was supposed to go. That's what I had planned."

And the pain of that will never, ever, ever, ever go away... because the loss of that dream is a very, very significant loss.

But... if you spend your life mourning the fact that you didn't get to Italy, you may never be free to enjoy the very special, the very lovely things ... about Holland.

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The table below lists the average age and range of milestones attained and should be used only as a guide and for reference.

Early Years

Most RTS children are born with a physical appearance different from their family background with features and characteristics being recognised by an experienced paediatrician/clinical geneticist.

This chapter describes characteristics and features that develop in the early years. Each child's development is unique and the following information covers the most apparent characteristics.

Nearly all parents face problems with feeding their child. During the first months, breast feeding can be difficult. The baby often drinks slowly and maybe vomits much of the feed and growth may fall below average as a result. As the child gets older there can be a tendency to become quite sturdy. Babies are prone to chest infections. They often have colds, which they can't quite shake off and this also can make feeding particularly difficult.

Gastro-oesophageal reflux and vomiting can be a major problem resulting in a

failure to thrive; vomiting can often be described as "projectile vomiting" when the entire feed is vomited with some force.

Congenital abnormalities of the heart appear in around a third of babies born with RTS. They have a variety of cardiac defects which are detected initially by a "murmur", which is simply an extra noise heard by a doctor listening to the baby's heart. The term "heart murmur" does not tell the doctor the exact problem, but simply that there is a noise that will probably require further investigation. This will either be organised by a paediatrician or by a more experienced doctor specialising in heart problems (Cardiologist). The cause of a murmur will vary with some being harmless and simply requiring further monitoring. Sometimes children do require surgery at a later date.

RTS individuals are notable for their friendly and cheerful character. They make contact easily and are very affectionate. Their life expectancy is normal and they continue to learn new skills throughout their life.

Skill	RTS Average (mths)	RTS Range (mths)	Normal Range (mths)
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

Common Characteristics

Skeletal

Short stature

Skin

Risk of developing keloids or excessive scar tissue resulting from trauma or surgical procedure.

Cardiac

There are thought to be congenital defects in approximately a third of people with RTS. Some are innocent heart murmurs requiring no treatment.

Speech

There is a small percentage for whom speech will not develop, but most will speak between the ages of two to four. Children often understand more than they express verbally. Speech and language therapy can play an important part.

Cryptorchidism in males

This means undescended testicles. Nearly always an operation is needed to correct this.

Cranial features

Microcephaly (small head size); prominent forehead, broad nasal bridge and deviated septum.

Facial features

Slightly malformed ears; beaked nose, a highly arched palate, downward slant of the eyes (downward slant towards the outer corners), heavy or highly arched eyebrows.

Dental

A typical deviation of the permanent teeth known as 'talon cusps' (little hooks on the underside of the incisors).

Eyes

Over 80% of people with RTS have some sort of abnormality. It could be tear duct obstruction causing sticky eyes, conjunctivitis, cataracts, glaucoma and/or problems.

Hands and feet features

Broad thumbs and first toes, at times the thumbs and/or toes being angulated. Corrective surgery is possible in severe cases. Can be prone to fungal infections of the nail beds, and ingrowing toenails.

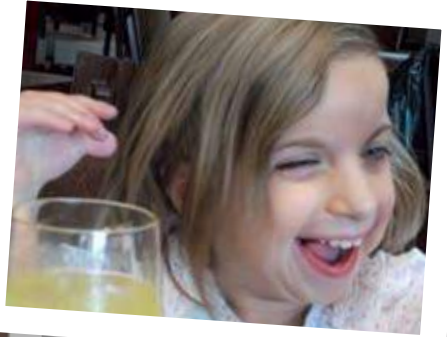


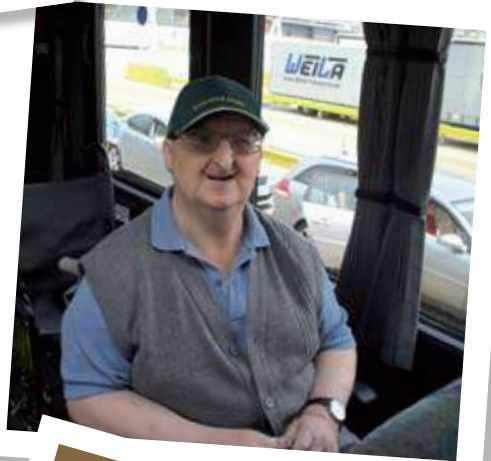
Photograph by the Garth Dawson Studio

0 - 5 years



6 years - Adult





Growing Up

The faces of children with RTS change quite a lot during the first years of their lives. For a timely diagnosis it can be important to know this.

Features often become more pronounced as the child grows older. At infant and toddler stage the face is often round and symmetrical. The bridge of the nose is somewhat more pronounced than is usual at this age. Towards the end of the toddler stage, the characteristic smile becomes recognisable. During school age, the facial features become more recognisable and are often described in literature as a 'typical RTS face'. The face becomes longer and is often slightly asymmetrical. The eyes slant downward often with heavy eyebrows and long eyelashes. The 'typical' laugh becomes quite recognisable.

The problems with eating and drinking which are fairly common in babies can last well into early childhood, but then they usually disappear. However, choking and vomiting remain weak points with many children. This can result in vomiting especially when laughing and/or coughing.

Motor development

The average child with RTS will sit and crawl between their first and second year. They usually learn to walk between two to four years. Their manner of walking

is rather wooden – often described as a 'Charlie Chaplin' walk.

Motor development can be encouraged by early physiotherapy. The physiotherapist can also be very important in the development of fine motor skills (pincer grip, brick-building, etc).

Mastering these skills will encourage some independence and the ability to get dressed, undressed, and feed oneself. Many RTS children require toilet supervision throughout development; however, some children are completely toilet trained by five years of age. In most cases it does not come until later, constipation complicating the process. They are often dry through the day, long before they are dry through the night.

Puberty

In most cases, the onset of puberty occurs around 12 years of age, which is similar to the rest of the population. In exceptional cases, this can occur as young as ten years old and also as late as 16 years old, or even around 30 years old. Young women begin to have menstrual cycles at the usual age as well. Some women can suffer from extreme blood loss during their period or from bleeding between periods. Taking the pill can help to regulate periods in these cases. It has been reported that two women with RTS, have had children. One woman had an unaffected child and a son born with RTS. The other gave birth to an unaffected child. We must then

assume that individuals with RTS are physically capable of having children. The chance that they then pass on the syndrome could be as high as 50%.

On the whole, adults with RTS are healthy and seldom ill with common ailments such as colds. Life expectancy is normal. A small number of adults never overcome their feeding problems. They continue to choke easily and vomit often.

There can be an increased need for sleep as they mature, this needs to be recognised, as if not, they can become bad tempered and difficult to get on with. A mid day nap is just the tonic. The majority of young people and adults can amuse themselves well: music, swimming, looking at books, puzzles, cycling, supervised horse riding and dancing remain favourite pastimes. They remain cheerful and still have a friendly disposition. Individuals should be encouraged to develop any interest they may have. Sport could improve hand/eye co-ordination, improve physical strength, and help combat any tendency to be overweight. Social contact could then be encouraged through these hobbies, as there is a tendency for individuals to isolate themselves as they get older.

“My daughter developed pubic and underarm hair at a particularly early age (about 9 if I remember correctly), and she developed mild acne and greasy hair around the same time.

Tests were done to see if it was due to early onset of puberty, but were pronounced negative. The levels of different hormones in her blood have also been tested and are apparently normal, but I do wonder about hormone imbalances: although her periods started at about the usual time, they have been extremely irregular – she once went a whole 12 months between periods - and she has male-pattern hirsutism (hairiness).”

Katie's Mum

“Philip was born in 1977, the youngest of our three sons. He had all the usual problems associated with RTS, poor feeding, constant vomiting and constipation, hence very slow weight gain. He was generally a very happy baby even though he suffered from constant chest infections and always liked to be carried around (it is amazing how quickly I learnt to do most jobs with one hand while Philip was in my other arm!).

The paediatrician said that he was suffering from ‘developmental delay’ it was not until he was almost five years old that RTS was diagnosed. When we read the information that was available and saw photographs of other children with the syndrome, we found it hard to believe that it had taken so long to reach a diagnosis; Philip was so typical of those with RTS. It may sound strange but it was actually a relief to have this diagnosis, we now knew that it was not our fault and felt we were no longer alone.



Philip

Around the age of 12-13 years, Philip became increasingly interested in pictures in the catalogue of ladies in swimsuits and of his own body changing, hair under his armpits and in the pubic area. He began to keep touching himself and we were constantly telling him to ‘get his hand out of his trousers’ and to ‘stop fiddling’. At school the teachers kept sending him to the toilet, no doubt thinking he needed to spend a penny, this became more of a problem when he was spending longer in the loo than in his classroom.

The school medical officer suggested we have some professional help as it seemed that this had become yet another of Philip’s obsessions.

A psychologist visited us at home on a number of occasions as well as seeing Philip at school. He suggested that

Philip was only to look at catalogues in his bedroom and every time that he touched his groin we ask him to go to his bedroom and draw the curtains if he wanted to 'Fiddle', saying "it isn't wrong but you just don't Fiddle anywhere else but in your bedroom". The teachers at school used the same phrase, and although it took quite a few months for Philip to stop touching his groin at inappropriate times and to go to his bedroom without being reminded when he wanted to 'Fiddle', it seems that being consistent and using the same words eventually showed results.

Philip has always had obsessions, they have often caused considerable problems, everything had to be done in the same way at the same time, his routines had to be adhered to or else he would become extremely agitated. When Philip was 22 years old we read a medical report on some research done in America where it was noted that 'obsessive compulsive disorder' is fairly common in those with RTS. I showed this report to Philip's consultant and she suggested he try some medication which she had known to be beneficial with this type of behaviour. Philip has been taking medication for a number of years now and we have all noticed a tremendous improvement in all aspects of his obsessive behaviour. I had always said that I would never put

Philip on drugs to alter his behaviour, perhaps believing that it was because I would then be admitting that I was not coping very well. How glad I am that I agreed to a three-month trial of this particular drug, the difference was quite remarkable.

Philip is now in a 'shared care' programme (in a residential home) with the view that this is where he will eventually make his permanent home, he has gradually built this up to only coming home alternate weekends and he seems very happy with this arrangement."

Philip's Mum and Dad



Philip

“Steven was in respite care and I was on my way to pick him up but I was running late so I rang to let them know I would be a few minutes late, they said that was fine as Steven was watching Coronation Street.



When I arrived the staff told me he had had a good stay but he had tripped over just after tea. He had picked himself up and they had asked whether he had hurt himself. He said No! When I walked into the room Steven was sat down on the settee watching the TV as he always did. He saw me and his immediate words were “stupid boots fell over”. As I was walking towards him I was checking him visually from head to toe and I asked him if he had hurt himself. He just grunted. By this time I had noticed he wasn't moving his right arm. As he moves his arms a lot when he talks, it was unusual and alarm bells were ringing in my head. I asked him if his arm was sore. He grunted again. He was wearing a jumper so I lifted the sleeve of the jumper and he had a clear dislocation of the elbow. The staff were mortified because they had checked him and he was acting as he normally would.

I drove Steven to the hospital. He never complained of pain all the way there and the hospital staff were amazed that he was not showing signs of pain. I explained that he had RTS and has a high pain threshold and he has been known to walk on undiagnosed fractures before now. The injury was not only a dislocation but a bad fracture and he needed 3 hours of surgery to put his elbow back together. Steven's change in the tone of his voice when he answered my questions was the only indication that he had hurt himself.

Now he lives in supported living and all the staff that support Steven are informed of his high pain threshold and if he does show changes in his behaviour to check he has not injured himself or is in pain due to illness.”

Steven's Mum

Growth

Individuals with RTS tend to be shorter than their peers throughout life. The average height in adult males is 5'5", while in females it is 4'10". Like all children, the height is variable and will also be affected by family background.

Boys tend to be overweight during school age years, often being described as stocky or sturdy in build. Girls may be overweight in adolescence. This can be a serious issue for some individuals.

Behaviour

Individuals with RTS are usually very happy and social people. They love attention and know no strangers. They are very friendly and cheerful and make contact easily in fact sometimes too easily, which can cause worries. At the same time, they can be stubborn. On the whole they are socially well developed and generally co-operative. They can be very easy going, but often find it difficult to concentrate for any length of time on one topic. When teaching them anything, lots of patience and perseverance will be needed. They work well with routine and are not fond of large or noisy groups.

A very common finding is that individuals with RTS have a very high pain tolerance. As they cannot articulate their pain, it is often only through behaviour change that you become aware that there is a problem.

See Appendix 1, "Behaviour in Rubinstein-Taybi Syndrome", for more detailed information.

RTS children love to touch everything and like to manipulate electronic appliances, buttons, knobs, dials. Many learn instantly how to work the video or DVD player, constantly finding and replaying their favourite bit! The obsession with opening and shutting all types of doors can be common; this is where cupboard locks come in handy! Many individuals with RTS exhibit autistic-like behaviours, ie. repetitive, compulsive or ritualistic behaviours, or unusual communication and social interactions.

Examples of this are rocking, spinning and hand-flapping in the early years (which they may grow out of), lining items such as toys up in rows, having to have things like patterned plates and bowls the "right" way round, and needing to touch certain things at particular times or in particular ways. All children with RTS continue to make progress and learn new skills throughout life. Each child is an individual, and will attain some milestones before they obtain others. Parents

The Life of a 32 Year Old

should try not to compare their child's progress with another child, although this can be difficult. spurts of progress and achievements will happen and when they do, it makes all the hard work worthwhile.

Learning should be fun. Singing instructions like "This is the way we brush our hair" whilst brushing the hair can help. Listening to tapes in the car and talking to your child all the time, even if they have no spoken language, will all be beneficial. An electric toothbrush for brushing the teeth and even a musical potty will all help to make learning more enjoyable for both you and the child. Although it has been said that these children work well within a routine, care should be taken to ensure that they still remain flexible and that they can cope with change.

Many parents have reported problems with the inflexibility of the children, they do not seem flexible in their views and they present great disappointment when things do not go their way, or they do not get what they want. They can then be very stubborn, sometimes resulting in temper tantrums, aggression and the refusal to compromise.

During adolescence, children with RTS, like other teenagers, enjoy their own space and time away from people. They often come home and go to their bedrooms to be alone, play music, watch TV or have their meals. Many react if this space is invaded, just like a normal growing child!

"Richard is now 32 years old. We gave him a special party on his 30th birthday, with over 70 guests and complete with a professional firework display. He still watches the DVD David (dad) put together for him and even as I write he is watching a DVD of a holiday in Florida, done in 1993!

Richard has a busy routine during a normal week. He goes to SACAR (For people with Autism and Asperger's Syndrome) on Mondays to help develop his social skills. He is still not very tolerant and hates queues, etc. He is also learning to stay calm and finds this easier to do if he thinks of hot air balloons. We went to a balloon festival in Bristol a few years ago and he watches the DVD which helps to calm him down.

Richard spends the rest of the week at the "Restaurant in the Church". He works in the kitchen and escorts his 'old dears' to and from the pensioners' lunch club. When he cooks he wears his chef's black and white check trousers, a 'sous chef' t-shirt and check hat. He enjoys responsibility and likes to think he has his own special job. His signature dish is parkin and he can whip up eight at a time all by himself, and these are good enough to sell at the church. Every now and then he goes for a



Richard

respite weekend with Julie and Charlie ('Shared Care' participants). Charlie is in a Billy Fury tribute band, fronted by Colin Gold. He was one of the original backing group members in the 60's. So when they play locally Richard attends and goes backstage afterwards.

Richard has another carer called Brett, who takes him out for a whole day once a month. This is funded by the Independent Living Fund (ILF). They travel far and wide as Richard likes to visit museums and country houses. Like most kids he will do things for others that he won't do for us. And all his carers think he's great. Sometimes I wonder if they've got the 'right' Richard.

He still sees a lot of things in black and white and if he makes up his mind not to go somewhere he won't. In June 2009, we went to Cyprus and he was with us when we chose a trip to go on a millionaire's boat for a day, including champagne, hot and cold buffet and which anchored in a bay to allow guests

to swim which he loves. However, come the morning of the trip he would not go. No amount of cajoling, promising, even threats and bribes - we tried it all. In the end David had to go alone - we wasted about 150 euros. We've decided 2 weeks on holiday is too long for Richard, he gets homesick. In future, we may have to take him for a week and think about respite for a week, while we venture somewhere on our own.

Richard spends a lot of time in his room watching DVD's, listening to music, etc. and would have all his meals there if we let him. He loves theatre, musicals, Harry Potter; Ken Dodd (we've been to his Christmas show 5 times), Blackadder; Open All Hours, to name a few. He has a wicked sense of humour and a great memory. He acts out musicals in his room and dances along. One day the ceiling will come down, especially dancing to 'old bamboo' from Chitty Chitty Bang Bang.

David is now retired and he has taken over from me and receives the Carer's Allowance. I now have a bit more freedom and can take on more hours at school. Less stress all round."

Richard's Mum and Dad

Speech and language development

Speech problems are present in about 90% of RTS children. Speech therapy using a Total Communication Approach should be started as early as possible. Speech is one of the slowest development areas of an RTS child and because a small percentage of children do not develop speech, signing should be considered initially as a form of communication. This can begin at a very early age within the first months of birth. Speech generally starts later than that of their peers, with children tending to say their first words between the age of two and four years. Sentences often start between the age of four and five years. The child can often understand much more than what they can verbally express which can lead to frustration and anger. It is important to look at other means of communication. This can be started at an early age, firstly through gestures/signals and then with the introduction of signs and/or symbols. The Makaton system is very commonly used.

The Makaton Vocabulary is a Language Programme which provides a basic means of communication and encourages language development for children and adults who have varying degrees of learning difficulty. Speech, signs and symbols are combined with the Makaton Vocabulary Language Programme. The signs used in the United Kingdom are from British Sign Language and have been selected and standardised for use with Makaton

Vocabulary. A wide variety of teaching materials are available and include – language programme manuals, line drawing illustrations of the signs, symbols glossary, record sheets, video tapes, picture cards etc. A research information service is available to keep Makaton users informed of current relevant research.

As a child's speech develops, signing may be dropped. Makaton will not hinder the speech, but will help to develop it.

Some children also have an articulation problem. Most have a high and narrow palate a small upper and lower jaw and a little mouth.

A speech therapist will not only help with speech problems and language development, but will also be able to give advice on feeding problems, which are connected with the deviant mouth motory. It is worth seeking advice for speech therapy as early as possible, as, depending on your local authority, there may be difficulties in securing regular speech therapy.

Education

The range of special educational needs is extremely wide, ranging from mild to severe learning difficulties.

In all the old RTS literature, children with RTS were most often described as having severe developmental delay. However, now we know the range is very wide.

Most RTS children will have some degree of developmental delay. Some of the children will do very well in a mainstream playgroup, nursery and school, but they will often benefit from having a support assistant. However, other children, whose development is progressing more slowly, cannot cope in mainstream placement and would benefit from the specialist care that a special school could offer. Parents should look at all options, talk to the staff about what they could offer the child and where necessary discuss it with a professional. An educational psychologist and/or a responsible officer will often help you make the right decision. Your child will probably require a statement of his/her educational needs. This is where all the people involved with your child prepare a report about your child's needs and progress. All the reports are considered, along with a report from you and then a statement will be issued. A review of this statement should take place every 12 months and as a parent you may need to work hard to ensure this actually happens. This will vary from area to area.



“When Katie left school at the age of 18, she went on to a residential special college. This means she lived there during term-time and came home for the holidays.

The course was 3 years long, and I think of it as her equivalent of going to university. She achieved a basic retail qualification, and there is a possibility she will be able to use it to find paid work. She also had great fun studying creative arts, and gaining more independence skills. She spent most of her leisure time at college in the social centre, and made some lovely friends. Being away from home was difficult for her, and for me at first, but we both got used to it, and I just felt it was the right thing for both of us in the long term.”

Katie's Mum

Young people

People with RTS will not lose the skills they have learnt and in most cases they will continue to make progress. Teachers and professionals should be made aware of this fact, as it could have an effect on the decision made about their future.

Most children have a Statement of Educational Needs and may have the option to stay in school until they are 19 years old and others leave school when they are 16 (this may vary from area to area). It is therefore important for a discussion to take place between them, their parents, social workers and careers officers to discuss what suitable options are available after leaving. This is called a transition meeting, and it is the responsibility of the child's school to arrange for this to be held in year 9. A transition plan should be produced, and this should be reviewed at a transition review meeting every following year until the child leaves. Options after school include a residential placement at a special school or college; a local further education college offering a special needs course aimed at developing personal, social and other life skills; or a work experience course/placement.

“Our son John attended Green Meadows School until he reached the age of nineteen. This school catered for a whole range of pupils from moderate learning difficulties to severe and profound. John was approximately four when he started at the school, at that time the school was named Tranfield Special School but it later merged with a school that it adjoined and shared the playing fields with.

This other school was called Two Dales and catered for pupils with severe and profound learning difficulties. After the amalgamation the school was renamed Green Meadows. John was happy at school and made many friends. After fifteen years he had become comfortable with pupils and teachers alike. He was happy there and well liked. Also Kath was content in the knowledge that he would be supported when he became unwell.

John did suffer with migraines and the school would bring him home when he was unwell. We therefore decided that she would not return to paid

employment, the reason being that if John became unwell, she would be at home to support him and care for him. At that time we were still coming to terms with raising a child with special needs and it was only just after receiving the diagnosis that John started to attend school.

When the time came for John to leave school, Kath and I were really concerned that he was leaving a safe environment and he was going to attend a college that catered for pupils with special needs which was situated across the other side of Leeds. We were concerned about the travelling and the different environment that John would be experiencing. Green Meadows had all the hall marks of a friendly supported school and it was only approximate five minutes away by car:

Although John would be travelling in a mini-bus the thought of him going across Leeds was always in our minds. John soon settled into the College and again made some friends there; some of them had attended Green Meadows School so this certainly helped John settling into the college.

Throughout the teenage years John would sometimes have some outbursts and teenage tantrums. He quickly became upset and sometimes very

emotional, which he still does.

It must be hard for people with special needs to communicate and voice their concerns and display their feelings. I think that was part of John's outbursts, which coincided with the door slamming!

Children in mainstream schools develop friends and some of them last for a life time. Most of John's friends were at school so consequently he did not see them after school time. He would "play" with children in the street but they were much younger than John.

For me, this is a downside for people with special needs, whether they are at school or college. They don't seem to have the consistency of friends, John had "friends" who were far younger than him and when they grew up they would interact with their peers. This left John again without any local friends.

Like all teenagers John likes music and watching films. The music can become a little monotonous; he plays the same discs and sings along with the words at such a volume!

Like all children and teenagers, they go through various stages and it's not all negative, we had many laughs, and still do with John. Many of them like sport and John is no exception. Football and rugby are his favourites. Tottenham

are his favourite team. I think that came about because of the "diving" around by Jurgen Klinsman a few years back. He has also met many people since he started to watch the Leeds Rhinos; he has got accustomed to the people who sit around us at the home matches.



John

We go into the player bar after the games and John knows some of the players and counts Rob Burrow, a star of the Rhinos, as one of his "Friends". When Rob was younger, he and John would wait outside the player's dressing rooms and try and get as many autographs as they could. Of course me being the protective parent, I would ask Rob to be careful and to wait for John, he wasn't as fast as Rob and I had visions of Rob running off and leaving John looking lost with autograph book in hand!

One of the positives that we have experienced is the group that John attends on a Tuesday night. The group is called People in Action and they cater for people with a disability. There are about six people who attend and John really enjoys it. The group visit the local bowling alley, snooker centre; they go walking to a pub; all the things that teenagers or young adults would do!

John speaks very positively about the group and he is really excited when the letter arrives to inform him of the next week's activity.

It can be difficult when RTS people are going through the teenage years, there are frustrations. I believe a little more patience is needed and try and remember that it's frustrating for them more than anything else. All children are different and people with RTS are the same, they need time and understanding when they are experiencing different stages in their lives. They are the same person, so go with the changes, and laugh with them. There will be tears, some upset but plenty of laughter. They still give their love and affection, despite their advancing years!"

John's Mum & Dad

What happens after leaving school?

As children with RTS grow up there are options to pursue including attending college, a day centre and/or working within a supervised and appropriate environment.

There are many voluntary organisations set up to provide opportunities for the personal development of people with a learning disability, offering day, short and long stay, holidays and assessments. The emphasis is to enable the person with a disability to enjoy an adult lifestyle which is full and socially valued, to enjoy security and a sense of performance.

“Maddie left school at 16 as the special school she was at didn't have post 16. If she had had the opportunity to stay on the things that she has since accessed would still be there and available, just a bit further down the line.

She attended Moulton College Northampton, from age 16-19 yrs, which we are fortunate to have virtually on our doorstep as it has an excellent reputation. She began with a course "Rural Skills" which was two years in duration. In the first year she covered many small modules, including Small Animal Care, Horse Care, Horticulture, Agriculture,

Countryside Management. Much of the work was practical and hands on but there was written work, which could be done on the computer or handwritten, usually a combination of both. There was a good ratio of staff to support these activities and classroom work.

The second year consisted of choosing one of these modules to concentrate on and complete work placements. Maddie was "encouraged" to opt for Horticulture as it was deemed easier to find work placements in this field, although her first choice was Agriculture as she enjoyed Sheep Dipping.



Maddie

A third year, "Bridge to Work", was optional, but students were carefully selected so as to ensure suitability. Maddie completed this as she didn't yet have the work skills to go on after college and find employment. Ability of students on these courses was quite varied, although they were grouped together in small appropriate groups. At the end of the third year, we had already lined up a day service which was aimed at young people no longer in education who needed to improve social & work skills. This was funded initially by the local council, but after it was agreed to extend the duration, we had to apply for funding to pay for this service and the provision of taxis as Maddie is unable to access public transport independently.

This service had various strategies in many aspects of key skills, numeracy, literacy, listening, working as part of a team etc. but was intended only to be short term and after about 9 months it became apparent to us that we needed to look elsewhere. We had already started researching but they allowed Maddie to extend her terms whilst we found something suitable.

Then came an Adult Learning Training Course which is run by our County Council for adults of any age who have a learning disability. This is called "Chefs in Training" and covers NVQ in Food & Hygiene skills, starting at Level 1. This takes one academic year to complete, and if successful can lead to a second year, Level 2, and potentially a third to do Level 3. Maddie is half way through her second year, and attends Mon, Tues & Weds, 9.15am until 3.15 pm, (Weds afternoon is optional and offers music, dance drama, arts & crafts). "Chefs" is term time only so there are quite a few holidays to take into account but it is funded by the local council; only the end of year exam has to be funded by the learner.

Since age 18yrs when Maddie was no longer supported by Children's Services, we were referred to Adult Services and so began the process of applying for funding for respite care, transport costs and anything else that required payment relating to Maddie's day support and access to services. This was by means of Direct Payment initially, but Northamptonshire was soon to be piloting a new service, Self Directed Support, funded by an individual budget. Soon

after the pilot scheme was successful and launched, we were one of the first families to apply. We now have an individual budget for Maddie's needs, which covers many things and has now allowed her social life to expand hugely.

We were very fortunate to come across Louise whom Maddie has known since college. The arrangement began by Louise taking Maddie out, then her staying overnight. They have been her respite family now for more than 3 years, and through them we have met Zoe and Caroline who support Maddie, mostly evenings but occasionally daytimes too. They have become friends for Maddie, and take her out during the weekday evenings, tenpin bowling, cinema, swimming, or just having a DVD night and some "girly time." This is all funded through Maddie's budget, for the benefit of the budget and relevant paperwork they are known as PAs (Personal Assistants). Louise and her family also take Maddie on holiday during the summer break, to the pantomime, theatre, pub and/or restaurant etc; whatever Maddie wants to do basically.

They have become a second family to her and, as a result, she has a wide circle of friends whom she texts and calls



Maddie

regularly from her mobile phone, credit for which has to be paid for out of her personal money and not the budget!

Maddie is now approaching her 22nd birthday, and enjoys a fantastic social life with people who are not her parents or siblings, quite right too!"

Maddie's Mum & Dad

Siblings

Having a sibling with RTS at times can be challenging and the siblings feel a range of emotions. Below are comments from siblings who have RTS brothers or sisters.

“I could not imagine a life without my brother Steven. The world would definitely be less colourful. As I was growing up this was hard to see because I felt like a victim because Steven was different, had greater needs to be met.

However all those feelings disappeared when you got a ‘Steven Hug’! I am convinced it could warm the coldest of hearts; and being around Steven made me feel like I was the luckiest person in the world.

Over the years we have developed our own little signs and nicknames which still remain today and probably for the future. Now Steven and I will continue with our lives independently of each other, but nothing will ever remove the bond that we have. I don’t think I could have asked for a better brother!”

Charlotte



Charlotte and Steven

“My sister Olivia is age 7 and has RTS. My name is Isabelle and I am 10 years old. We have a little sister called Lily she is 3.

We live by the seaside in Rustington West Sussex. I was 2 years 9 months when she was born, we found out that she had RTS. Although she's 7 she acts like a cheeky toddler.

She gets a taxi to a special school. Where they go out on the bus a lot for trips! Unlike me working hard at school. Although I do get to go to siblings group (sibs R us)/young carers and RTS events The most annoying thing is having to play duck duck goose all the time. But all in all she's good fun!

But she's not very good at potty training and she's nearly there but still need's help getting dressed (same with a few people I think).

More about siblings can be found in:
Appendix 2: “What is it like to have a brother or sister with RTS?”
Appendix 3: “The Experiences of the Brothers and Sisters of Children with Rubinstein-Taybi Syndrome”



Isabelle and Olivia

Sometimes when I try to make her laugh she laughs or smacks me or Lily but I don't know why? That all adds up to getting my attention and understanding her. I am waiting for the next RTS event to talk to people who have read my article and have brothers or sisters like my sister Olivia.”

Isabelle

Medical facts, hints and tips

The medical points in this chapter will not apply to all individuals with RTS. They may have some, all or none of these problems. The degree in which they can affect a person will also be variable. All concerns should be discussed with your doctor or health visitor. See also 'Anaesthetic' section.

Feeding

A relatively common problem in babies and young children with RTS is feeding, particularly breast feeding. It is difficult for them to master sucking and swallowing; the baby often drinks slowly and then brings up much of the feed. As a result, poor weight gain tends to be noticed. Projectile vomiting, where whole feeds are brought up with force, also affect weight. Gastro-oesophageal reflux can also occur. Babies are prone to bronchial infections and colds that can aggravate the feeding problems. Babies usually grow out of these problems in the first year or so, but in severe cases medical intervention is required. Any concerns should be discussed with your doctor or paediatrician. Thickening of feeds; starting early with solids; position during and after feeding; and sometimes medication are all options.

Weight Problems

There is a common tendency to be overweight, which can be at an early age or come on later, perhaps at puberty. The way to manage this is by healthy eating and exercise, there is nothing specific to RTS. However some RTS people

have a great interest in food which may contribute to this problem.

Constipation

Severe constipation can be a major problem for children and adults. A high fibre diet, laxatives, suppositories and enemas often have to be used. Sometimes diarrhoea can also be the problem.

Eye Problems

An eye specialist should examine all children as more than 80% will have eye problems. These can include squinting, cataracts and blocked tear ducts. A blocked tear duct can lead to very sticky eyes; sometimes they are unable to open them after sleep. In some babies or young children, glaucoma can occur. This is an increased pressure in the eyeball, which if left untreated can cause serious problems with vision. By the time the children start school, as many as half will require glasses.

Ear Problems

About 50% of children suffer from recurrent ear infections (glue ear). This can mean a temporary loss of hearing.

In the first years, colds and bronchial infections are also very common. Grommets and sometimes the removal of tonsils and adenoids can help. You should be referred to an ear, nose and throat specialist (ENT), if any of these problems are found. Often this clears up between the fifth and tenth year of age.

Undescended Testes

Males with RTS usually have undescended testes (UDT). Nearly always, an operation is needed to correct this. Ideally, this should be done at an early age, before the age of two. The purpose of the surgery is to bring the testicles into the scrotum, which is the normal environment and is usually two to three degrees cooler. There is an increased risk of tumour or cancer developing in children with UDT and testes that are not corrected until the teen years, and this is best treated with the removal of the testis.

Kidney Problems

Minor structural abnormalities of the kidney have been reported. There is limited material available in this area, but it would be worthwhile screening people with a renal ultra sound to have a close look at the bladder and kidneys if there are concerns in this area. Sometimes urine can reflux from the bladder to the kidney when urinating.

Heart Problems

About a third of the children have heart problems. There may be a variety of defects, none of which are specific to RTS. Sometimes an operation is necessary. See 'Early Years' Section. A cardiac evaluation is recommended for all individuals with RTS.

Hands & Feet

Nearly all people with the syndrome have broad thumbs and broad big toes. Tips of the other fingers may also be broader than usual. In some individuals the thumbs may also be angulated, often described as 'hitch hikers thumbs'. Surgical correction may then be beneficial to improve their use. Where the big toes are angulated, surgery may also be of benefit in order to find suitably-fitting footwear. Shoe supports may be necessary to counteract flat-footedness.

It should also be noted that individuals may be susceptible to fungus infections of the fingernails and toenails and ingrown nails. Webbed feet and occasionally an extra toe may feature.

Seizures or Epilepsy

Some children will be affected by epilepsy. The seizures are well controlled through medication.

Sleep Apnoea

Children may experience sleep apnoea. This is when the breathing is laboured and obstructed at night, resulting in a drop in oxygen levels. This is sometimes associated with enlarged tonsils/adenoids. An anaesthetist should be made aware of this possible problem. See 'Anaesthetic' section.

Keloid Formation

In a number of children and adults there is tendency for keloid formation. This is excessive scarring or a raised scar. This most commonly occurs on the chest but may also be seen on the arms and back. In some people it seems to happen spontaneously and in others it happens following any type of injury which causes inflammation of the skin. This could be a surgical or accidental cut, a pierced ear, acne or a graze caused by a fall. A cure for keloids is not yet available but there are now treatments with dressings which can help.

Dental Problems

In about 65% of the children there are dental problems. As mentioned before, most children have a high and narrow palate, a small upper and lower jaw and a little mouth. Because of the mouth structure, there is less room for the teeth and therefore they are closer together and can be more difficult to clean. A small mouth opening can also make brushing more difficult. An electric toothbrush is easier to use and many children also like the sensation.



(Photograph by the Garth Dawson Studio)

A high percentage of older children and adults have talon cusps on the underside of the permanent incisors (a pointed bulge which may contain pulp). In the hollow between the tooth and the bulge, tooth decay can form or the point can irritate and damage the tongue making treatment necessary. Talon cusps usually occur only in second/permanent teeth. Many older children require some orthodontic treatment, but orthodontic involvement is recommended from an early age. Selective extraction is also another form of correction for overcrowded teeth. Where there are heart defects, antibiotic cover may be needed for dental treatment.

Orthopaedic Problems

Besides the possible problems with the thumbs and toes, other orthopaedic problems could include dislocated knee caps (patella) and curvature of the spine (scoliosis). Every child with curvature of the spine should be examined regularly, especially in the beginning of adolescence. The degree of the curvature can increase severely

when there is a rapid increase in growth. Physiotherapy exercises, swimming and horse riding may all be beneficial in strengthening the back.

Hypotonia – this is poor muscle tone, resulting in general floppiness and weakness. It can delay motor development i.e. sitting, standing, and walking. Hypotonia in the hands can affect fine motor control, causing difficulties with buttons, pens and pencils, shoe laces etc. Hyper-extensible joints – this is an unusual, excessive degree of movement in the joints. It seems to affect the elbows, knees and ankles in particular. A combination of the above could partly explain the wooden manner of walking. Support for the ankle area is sometimes necessary in order to prevent injury resulting from weakness in that joint.

Perthes disease of the hip joints can occur, mostly between fifth and fifteenth year. This disease can make walking both difficult and painful. Treatment usually consists of providing supporting appliances (braces). Sometimes an operation is necessary.

Anaesthetic Problems

In a number of children with RTS anaesthetics can cause problems. A muscle relaxant can cause the sides of the pharynx to collapse against each other making the insertion of a tube for breathing very difficult. An endotracheal tube should be inserted for

general anaesthesia to avoid aspiration. Anaesthetists should always be made aware that this could be a problem beforehand, even if there has not been a problem previously. It is also wise to draw the attention of the family doctor and paediatrician to this point so they are prepared for any acute situations. Due to lax ligaments, careful positioning of the neck must also be observed. Some individuals can have other problems whilst under general anaesthesia, such as altered cardiac rates and rhythms causing arrhythmia. Again, surgeons should be made aware of this possible problem.

Should my child be immunised?

In one study group 400 immunisations were given. The overall complication rate was 5%. This is no greater than for the rest of the population. Reactions consisted of a mild fever and irritability. Therefore there seems no reason to withhold immunisations for RTS children. Immunisations are important in order to prevent certain serious infectious diseases.

You should discuss any worries with your GP or health visitor.

Activities

Many of the children enjoy water sports and swimming. They enjoy horse riding (usually assisted) which can also help balance and reinforce positive behaviour. They love books and watching television.

Playing outside on bikes, scooters, swings and slides can occupy many a pleasant day. Sandpits and even soil can be a great source of pleasure. They are very responsive to any form of music and many parents have found music therapy a lifeline for education and also for working on behaviour and social skills. Children can develop well when working to a routine and in 1-1 or small group situations.

As they mature, they may find pleasure in things many other people do, according to their ability and preference. Inclusive youth clubs, dance classes and Brownie/Guide groups; visits to the cinema/theatre, ten-pin bowling, eating out or meeting up with friends are all possibilities. If there is a weight problem, it may also be desirable to encourage plenty of physical activity (where possible), such as walking the dog, swimming, or playing football.





Independence

The majority of people with RTS will require some form of support to achieve any degree of independence.

The Social Services department of the local authority can help them towards independence by making an assessment of their requirements.

There is currently a move towards “person-centred” planning, where, after the assessment has been made, a plan is formed as to how the individual's wishes and needs will be met. If the services run by the local-authority-are not appropriate, then others (e.g. a personal assistant) can be bought in, and paid for by what are known as direct payments. Some local authorities have gone a step further and create an individual budget out of which the service-user (with assistance if necessary) has to pay for all the activities and services they need. The government wants this system to be extended to everyone eventually.

Sarah



“For the past 19 years, our daughter, now 38, has been looked after by a Kent charity. Needing care and supervision 24/7, she lives with seven others in a purpose-built house in a small town.

The trust has a rare-breeds farm, ancient woodland, and opportunities for horticulture, art, craft, music (her favourite), IT training and life skills projects. Social events take place at the farm and medical needs are met locally. As we live 75 miles away, she visits us for weekends, or a bit longer, about 10 times a year, which seems to work very well, and we can keep in touch by telephone in between.”

Sarah's Mum & Dad

“Jenny has been in supported living for 2 years. Jenny lives with another lady in a semi detached house with 2 young men living next door.

The house is rented from a housing association, and the carers are employed by a private agency. Social Services allocate how many hours of support each person needs, a carer sleeps in the spare bedroom linked to the young men next door. At the moment Jenny gets 4 hours of one to one support.

She does 16 hours voluntary work at a local children's centre, also approximately 4 hrs at the pensions advice centre. She also is doing a computer course, plus trying her hand at washing, ironing, cleaning and shopping.”

Jenny's Mum & Dad



Jenny

Benefits

A person with RTS and their carers may be entitled to a number of social security benefits.

In England, Scotland and Wales, these are administered by the Department for Work and Pensions (DWP), and in Northern Ireland by the Department for Social Development (DSDNI) . Benefits are subject to change from time to time and so applicants are advised to contact the relevant department for the latest information on eligibility and how to make a claim.

Making a claim can sometimes seem daunting, but the government Benefit Enquiry Line, tel: 0800 882 200, and Disability Benefits Helpline, tel: 08457 123 456 are there to help. There are other organisations such as;

- * The Social Care and Health department of your Local Authority
- * Citizens Advice Bureaux
- * Welfare Rights Advisory Services
- * Contact a Family

who can also offer advice and assistance.

NB: This information applies in the UK only, and different provisions will apply or may not exist at all in other countries.



Contact Info

For free extra copies of this booklet or further information, to become a member of the Support Group, make a donation or discuss possibly fundraising, please contact:

Rubinstein-Taybi Syndrome Support Group

email: info@rtsuk.org.uk

Free extra copies of this booklet can also be downloaded from www.rtsuk.org/downloads.



Appendix 1

Behaviour in Rubinstein-Taybi Syndrome

By Laurie Powis, Jane Waite & Professor Chris Oliver
(The Cerebra Centre for Neurodevelopmental Disorders)

Social Behaviour and Social Understanding



“People with RTS are happy, loving, friendly individuals who love adult attention.”

People with Rubinstein-Taybi Syndrome are frequently described as happy, loving and friendly individuals who love adult attention¹. Research studies have shown that compared to other individuals with intellectual disability, people with RTS are more likely to want to engage in social interaction². Furthermore, research also highlights their abilities to play with others, make good eye contact, accept physical contact from others and show good social communication skills.^{3, 4} However, despite these findings it has been noted that some individuals with RTS can show a different social profile - preferring to spend time alone rather than with others.

Recently, the Cerebra Centre conducted a study investigating ‘social understanding’ in the syndrome. Social understanding was assessed using tasks that explored something that researchers call ‘Theory of Mind’. Theory of Mind involves being able to think about somebody else’s thoughts, feelings, beliefs and intentions. Researchers at the Centre found that some of the early Theory of Mind skills that require motivation for social contact are a relative strength in the syndrome – individuals showed a level of understanding that allowed them to help and cooperate with another person during different games. However, later developing Theory of Mind skills appear to be more difficult. This means that some individuals with RTS may find it difficult to put themselves in ‘somebody else’s shoes’ and think about what that person is thinking or feeling.



“People with RTS are motivated to help and co-operate with others”

Repetitive Behaviour

The term 'repetitive behaviour' is an umbrella term that describes some of the different types of behaviours that are often seen in people with RTS. These behaviours include the tendency to show repetitive speech and repetitive questioning. For instance, some parents report that the person they care for will 'get stuck in a loop' asking the same question or repeating the same thing again and again.

Some people with RTS are also known to show stereotyped movements such as rocking, spinning and hand flapping^{4, 5}. Parents have described behaviours such as 'flaps arms/ hands when excited' and 'makes fast movements with fingers and hands'³. These behaviours seem to be more common in younger individuals and end to decrease gradually with age.



"Parents have described behaviours such as "flap arms/hands when excited". However, these behaviours seem to decrease with age"

It has also been reported that around three quarters of children with RTS show behaviours such as 'insistence on sameness' and 'adherence to routine'^{4, 5}. This can mean that some people with RTS will prefer predictable routines, or may get distressed or angry when their routines change.

The Cerebra Centre conducted a study investigating what might cause repetitive behaviours and found that people with RTS who engage in repetitive behaviours, particularly repetitive questioning, may have poorer executive functioning than those who don't engage in repetitive behaviours. Executive function refers to brain processes that allow us to control and regulate our behaviour. These processes include inhibition (the ability to stop a behaviour once it has started), working memory (the ability to hold things in memory for a task at hand), and task switching (the ability to move easily from one task to another).

Encouragingly, the results of the study also suggest that people who are older are more likely to have better executive functioning and that many repetitive behaviours, including repetitive speech, are less common in older people with



“Executive functions are brain processes that help us control and regulate our behaviour. These appear to get better as people with RTS get older”

Other Behaviours and Changes with Age.

In addition to the social behaviour and repetitive behaviours described above, other behaviours have also been mentioned in the literature. Some studies have suggested that stubbornness, impulsivity, hyperactivity and sleeping difficulties may be common^{2,4,5,6,7}. Other studies have described individuals with RTS to be more emotional, excitable, show a short attention span and show a dislike for loud noises^{2,5}. Interestingly, research with other syndromes and disorders has linked stubbornness, impulsivity and hyperactivity to difficulties with executive functioning^{8,9}. However more research is required to determine whether the same processes lead to these same difficulties in RTS.

Although the majority of descriptions describe children as very happy and friendly, it has been suggested that people with RTS may show some behaviour change with age. Reports suggest that increased mood swings, temper tantrums and aggressive outbursts have sometimes been seen in early adulthood¹⁰. However, more

research is required to examine if, when and why these behaviour changes occur.

Summary

Research into the behaviours shown by people with Rubinstein-Taybi Syndrome is growing and, as outlined above, appears to suggest that there are certain behavioural characteristics that are common to the syndrome. However, it must be remembered that each child, teenager and adult is different and although some similarities can be seen across people with RTS, each person will display their own likes, dislikes, strengths, difficulties, and sense of humour.



“Although some similarities can be seen across people with RTS, each person will display their very own likes, dislikes, strengths, difficulties and sense of humour”

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Appendix 2

“What is it like to have a brother or sister with RTS?” By Dr Karen Aldred, D. Clin. Psychol., BSC

What is it like to have a sibling with RTS? Well my sister Sue has RTS so I should know! Or should I? The thing is, just as every person with RTS is different so is every sibling's experience. How it is for me isn't necessarily how it is for everybody. Sue was born when I was three, so I can't remember much before she came along. The things you grow up with tend to feel normal even if they might not seem so to others. So for me having a sibling with RTS seems normal and having a sibling without RTS would seem quite strange!

As well as having a sister with RTS I also happen to be a clinical psychologist by profession. I have learned something from my professional training and my day to day job about how people are affected by having someone with a disability in the family. In this article I am going to tell you what I have learned from speaking to siblings of people with learning disabilities and from looking at the research that has been done about siblings. I will also tell you about Sue, as I can't resist!

Pride and Joy

I remember how excited I was when my sister was born, and the anticipation I felt

on the way to the hospital to meet her; a little bundle of joy with loads of jet black hair. I was very proud, and that feeling of pride has never left me as I have watched my sister grow, learn to walk, talk, swim, learn to read and do all sorts of things that might be easy to others but have been a real challenge for Sue.



I am proud to be her sister. There is never a dull moment with Sue. She has the most wonderful sense of humour and is great fun to be around. Woe betide you if you try to tease her because she will always have a witty comeback, even if it is only putting her thumb to her nose and wiggling her fingers!



For siblings of children with learning disabilities this feeling of pride is common. Watching at close quarters as their siblings struggle to master things that others find easy gives us a real sense of how important and hard fought these small achievements are.

A couple of researchers interviewed children who are siblings of people with learning disabilities and found that they tend to have heightened empathy skills compared to other children. In other words they are very good at being able to put themselves into other people's shoes. This gives them a greater understanding of people in adverse circumstances. It is also a very useful life skill and helps people make good relationships in their work and social life.

Younger Siblings

The experience of having a sibling with RTS will be different depending upon their position in the family. For younger siblings, they are at first unlikely to understand that there is anything different about their siblings. Their understanding will increase as they grow older. Younger siblings have the rather unique experience of catching up with and overtaking the sibling.

Older Siblings

For older siblings the birth of a younger brother or sister is usually a major change to their life, whether or not the sibling turns out to have a disability. Excitement, a feeling of responsibility, being the 'big girl' or 'big boy', sharing your parents' affection and attention and your toys; these are common experiences for children when a younger brother or sister comes along. When the older child finds out there is something different about their sibling this can have an impact. The younger sibling is likely to grow up always knowing about the disability whereas the older child may find out more suddenly.

What is Normal Behaviour towards a Brother or Sister with RTS?

All siblings fight, squabble, tease and are jealous of each other. They try to pass the blame to one another for things going wrong. This is still true for siblings where one of them has RTS. The non-RTS child may be given more responsibility for these events just as usually older siblings are. But when you have a sibling with RTS you may feel more guilty after fighting or squabbling with your sibling. You may berate yourself feeling that it is not right to do this with someone with a disability (and if you don't chances are your parents will!)

Wishes and Sadness

Psychologists and other researchers have become very aware of the emotional reactions that parents have when they realise their child has a disability. They recognise that parents can be shocked, worried and go through a grieving process for the more able child they perhaps hoped for.

More recently their attentions have focused upon the feelings of siblings. In fact, siblings may go through a similar series of emotions to their parents. In essence they are grieving for the 'lost' abilities of the child. This is in terms of:

- a) what the child is not able to do at the current time (and all the repercussions this then has on giving time and support, hospital visits etc)
- b) change in expectations and hopes for their future (work, marriage, children etc).

These feelings of grief are quite normal emotions and are described elsewhere in this booklet.

I myself remember having daydreams as a child that one day they would find a 'cure' for Susan and that she would be able to play board games with me or come for a bike ride. Even as an adult it is hard not to occasionally think, 'wouldn't it be nice' if she was more able to do this or that.

In the Shadows

Because children with RTS need so much care and support, the other children in the family will often inevitably have to take a back seat. Their parents may have less time and energy for them than they would like. Often in families where there is a disability, money may be tight; either because of a parent needing to stop work to look after the RTS child, or because of saving and planning for the RTS child's future. This can put a strain on the whole family, including siblings who may have to make do with less.

Because of the needs of the RTS child sometimes it may not be as convenient for children to have their friends round or it may be that parents are not as able to ferry their other children to their friend's house or to other social activities. Sometimes a sibling may be the RTS child's main companion and parents may want them to stay at home and play with their brother and sister. All of this means that sometimes it can be harder for the sibling to make and keep friends and they may become a little bit isolated from their peers.

Sometimes children who have RTS siblings may experience people knowing them only as a result of their sibling and may get fed up with being known as, Tom's Brother or Mary's sister rather than by their own name.



Being Different

Children may feel embarrassed by how their sibling acts or looks and worry about going out and about with them. They may not be sure about how to explain RTS to their friends. Sometimes children may get teased or bullied as a result of having the sibling. On the other hand some children enjoy the feeling of being different or special by having an RTS sibling.

For me Susan was often a great ice-breaker when meeting new friends. She was much less shy than I was then and could be guaranteed to do or say something that would make us all laugh or get us all talking.





Responsibility and Pressures

For siblings there is often a feeling of needing to look after and protect the person with RTS. Children may worry about their RTS sibling being picked on or teased by other children. They may worry about how their brother or sister is going to cope with life. Sometimes children may have misunderstandings about what it means to have RTS. They may feel worried about asking their parents about what RTS means.

Children may also worry about how their parents are coping. Sometimes children end up in the role of an extra parent as there is so much to be done for their sibling. Sometimes they might feel that they cannot tell their parents about their own problems for fear of overburdening them.

Some children become the target of all the wishes and hopes that the parent had for the other sibling. In other words the pressure to achieve at school and at sport or other activities is heightened as all the expectations are upon them rather than shared. The parents' thwarted hopes and ambitions for the RTS child become directed into their other children.

Adult siblings may worry about what is to happen when parents are not around to care for their brother and sister or may already be providing some of the care. This responsibility can affect life

in all sorts of ways from deciding where to live to choice of husband or wife. My husband passed the RTS compatibility test early on in our relationship. Luckily for me he has welcomed Sue with open arms and they have lovely banter together.

Some siblings can feel pressurised by their parents' expectations of them caring for their sibling when they are no longer able. The sibling may feel guilty for example about not wanting to look after their RTS brother and sister in their own home. For myself, I have great respect for how my parents have given so much of their lives to caring for Susan and have done such a fantastic job of taking care of us both. I feel I have a lot to live up to.

Does having an RTS Sibling affect People Psychologically?

In 2001 researchers reviewed twenty-five different research studies on siblings with learning disabilities and found that siblings of children with learning disabilities were slightly more susceptible to negative behaviours/emotions, in particular depression. This does not mean that every sibling of someone with learning disabilities gets depressed, but just that they are slightly more likely to have depression than the average person.

Some studies have indicated that siblings of disabled children may show slightly more behaviour problems than other children . However other studies have not found any differences between the behaviour of these children and the general population .

There is some evidence that having a sibling with a learning disability can have some psychological benefits . Children learn to empathise quicker and can build up coping skills which help them to deal with life's ups and downs.

The ways in which parents support, and help their children can make being a sibling of someone with a disability a more positive experience.

I know for sure that I would have been very different if I hadn't had my sister Sue. For a start I would know far less about Knight Rider and Karen Carpenter (some of my sister's favourite conversation topics.) Who knows, perhaps I wouldn't have become a Clinical Psychologist. At the end of the day, I wouldn't trade my little sister for any other".

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Experiences of the Brothers and Sisters of Children with Rubinstein-Taybi Syndrome

By Jane Waite, Laurie Powis and Professor Chris Oliver

Being a sibling of a child with an intellectual disability has often been assumed to be a negative experience, particularly by people who have not had the experience themselves. However, the majority of published research suggests that, for most children, this assumption is wrong. Very few differences have been found between siblings of children with intellectual disabilities and other children, and when differences are found these differences usually favour the siblings of children with intellectual disabilities¹. Thus, although there may be some challenges for siblings of children with intellectual disabilities, there are also many positive benefits.

“He can make you smile even when you are feeling really, really rubbish. He gives you the most amazing hugs.”

Charlotte speaking about her brother Steven

Last year, members of Professor Chris Oliver’s team from the Cerebra Centre for Neurodevelopmental Disorder asked siblings of children with Rubinstein-Taybi Syndrome to tell them about their experiences. The siblings were keen to share their memories, thoughts and feelings and we have included quotes from siblings throughout this article. This article also highlights current research findings on siblings of children with intellectual disabilities.

“He makes me proud because he is very good on the computer. He has learnt to get onto CBeebies and he has learnt to play the games now!”

Brody speaking about his brother Karl

“He is funny, he sneaks into the fridge sometimes, and when it is quiet you know he is up to something!”

Mia speaking about her cousin Karl

Research from the 1990s has found overwhelmingly that siblings of children with intellectual disabilities are no more likely than other children to think badly about themselves, or have lowered confidence in their abilities¹. Furthermore, numerous research studies have failed to find differences in the number of behaviour problems exhibited by siblings of children with intellectual disability in comparison to other children. In the small number of research studies that did find more behaviour problems in siblings of children with intellectual disabilities, the behaviour problems exhibited by siblings did not place them in a clinically significant range, in other words, their problems weren't very severe¹.

“One difficulty with having a sibling with RTS was that when I was younger my brother did have problems and he needed a lot of attention to help him with his difficulties - so being jealous was the main difficulty for me. “

Charlotte speaking about her brother Steven

Interestingly, some studies have suggested that siblings of children with intellectual disabilities are more likely to have an 'internal locus of control' when compared with other children. An internal

locus of control means that a person feels that they are in control of their own destiny - it is the opposite of believing that your fate is determined by external forces. Children who have an internal locus of control are likely to be happier and less stressed. It has been argued by the researchers Burton and Parks, that siblings of children with intellectual disabilities “find psychological strength” from growing up with a brother or sister who has a disability².

“Some of my friends at school understand, but some may take the mick, but you can't really do anything about that. I know that I love them, and I like treating them just like any kids that we've adopted. Some people take the mick, but they don't understand” – David speaking about his two adopted brothers James and Ben

Researchers have also noted that the siblings of children with intellectual disabilities report less conflict with their brothers and sisters than other children^{3, 4, 5}. Fewer conflicts have also been reported by the parents and carers of the siblings. The majority of siblings appear to have positive, nurturing and

satisfying relationships with their brothers or sisters. However, it has been noted that often the quality and success of a sibling interaction with their brother or sister will depend largely on the sibling's ability to select appropriate activities that both they and their brother or sister can engage in^{1, 6, 7}.

“I like it when he does like new things. It made me happy because I taught him how to ride a tricycle - my mum couldn't do it, and my dad couldn't do it, but I helped him do it”

Daisy speaking about her brother Henry

“I'm proud of him because he does sign language and he is really good, and he is progressing lots”

Mia speaking about her cousin Karl

Siblings of children with intellectual disabilities may also assume more varied and dominant roles within the home in comparison to other children. These roles may include helping, teaching and managing their brothers and sisters^{1,8}. This can be beneficial for siblings of

children with intellectual disabilities because it leads to them spending large amounts of time bonding with their brothers and sisters.

“I help her with drawing, putting her shoes on, and with her coat. I also put DVDs on for her.”

Isabel speaking about her sister Olivia

Furthermore, engaging in rich, elaborate roles may have developmental benefits for siblings¹. It has been suggested that taking on these roles may enhance a sibling's ability to see the world from other people's points of view and understand the feelings of others. This suggestion has been supported by a research study that indicated that siblings of children with intellectual disabilities reported greater empathy⁸.

“I’ve just started a new job and we had to do lots of role plays for the training. Everyone kept coming up to me and telling me that I appeared really, really calm and really understanding and quiet. I think it’s because of him that I’m the person I am, but you don’t really notice it until someone tells you.”

Charlotte speaking about her brother Steven

In summary, although there may be challenges to having a sibling with an intellectual disability, the quotes in this article demonstrate that siblings frequently report positive aspects as well. It is also encouraging that the research literature suggests siblings may benefit

“I think he is happiest when he is eating his chocolate mousse, or around my nanna, or wearing his red trainers!”

Daisy speaking about her brother Henry



“Have patience, and when your brothers and sisters go in a little mood, be with them and help them, and don’t get frustrated even if they do bad stuff, because at the end of the day, they are still your brothers and sisters and still in your family, so you just have to help them along and still care for them.”

David speaking about his brothers James and Benn

“Keep on trying to teach your brothers and sisters new life skills, help them do their work and grow up to be healthy”

Matthew speaking about his cousins Ben and James

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