

A Personal Account of My Experiences With Rothmund-Thomson Syndrome (RTS)

Introduction: I am 33 years old and married with 2 children aged 8 and 5 years, went to mainstream school and now work part-time as an Executive Administrator for the Court Service.

I was diagnosed with RTS around 2 years old by Dr Charles Darley at a Dermatology Conference at the Royal London Hospital, Whitechapel.

At the age of 11, I was diagnosed with Osteosarcoma in my right fibula. At the time, this was denied to be anything with RTS, later finding out that it was connected. After a battling 2 years, I was given the all clear, however, had the fibula removed with a plate and screws and a height difference between legs, I have mobility problems, pain and swelling. I am awaiting the completion of specially made orthopaedic shoes to assist with supporting my walking and to slow down damage.

Skin and Hair

- ✚ Poikiloderma rash which fortunately has lessened substantially over time.
- ✚ Sun sensitivity
- ✚ Frequent cracking/cornified skin (particularly on lips, hands and feet).
- ✚ Frequent skin infections (particularly around ears, tummy button and groin).
- ✚ Café au lait patching on skin.
- ✚ Last year, loosing hair around the crown and premature greying!
- ✚ Sparse eyelashes and eyebrows
- ✚ Onychomycosis (Fungal nail infections)

Mother's Pregnancy/My Infancy

- ✚ Small for dates
- ✚ Delivered by caesarean
- ✚ Feeding difficulties so challenging weight maintenance
- ✚ Poikladerma caused challenges for poor Mum with being accused of 'putting me in front of the fire'.
- ✚ Dysplasia - broke ankles a few times for no apparent reason.

Ears, Nose, Throat and Teeth

- ✚ Mild hearing difficulties with several grommet and adenoid operations.
- ✚ Speech problems and therapy as a child. Mild speech problems - I find it hard to project my voice to be heard and have to speak slowly otherwise prone to stuttering.
- ✚ Acute Rhinosinusitus treated with antral washout
- ✚ Constant catarrh in nose and chest
- ✚ Bleeding gums
- ✚ Regular mouth ulcers
- ✚ Painful jaw
- ✚ Dry eyes
- ✚ Mild Asthma

Gynaecological

- ✚ Irregular and sometimes painful periods
- ✚ Irregular smears
- ✚ Polycystic Ovary Syndrome
- ✚ Easily put on weight
- ✚ Children were both born by elective c-section

Skeletal / Muscular

- ✚ Small stature however was taller at 4 ft 11.5 inches then my predicted 4 ft 10!
- ✚ Frequent headaches and neck/joint pain and stiffness which worse in cold and wet weather.
- ✚ Feeling lethargic
- ✚ Loose bowel movements
- ✚ Extremely boney Coccyx - painful to sit for long periods!

Medication (* current)

- ✚ Amitriptyline*
- ✚ Colecalciferol 800 (Vitamin D)*
- ✚ Fluticasone and Salbutamol inhaler*
- ✚ Dermal 500 lotion and Oilatum*
- ✚ Ketoconazole 2% shampoo
- ✚ Trimovate
- ✚ Spironolactone 100mg
- ✚ Minoxidil 2% Solution
- ✚ Spironolactone 100 mg

Summary of the last two years:

January 2012, had Atypical Pneumonia.

Constant leg pain and swelling as a result of poor mobility. Headaches and sometimes accompanying nausea, neck pain, stiffness, disturbed sleeping pattern, muscle twitches, constant fatigue, memory problems and poor concentration, pins and needles in hands and feet, frequently loose bowel movements, feeling very low, poor circulation. Hair loss and going prematurely grey.