Birmingham Women's and Children's NHS Foundation Trust

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Please quote our reference on all correspondence: 62631.0

Dictated Date: 23/06/2023
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Parent / Guardian of Miss Kerry Horner
19 Greendock Street
Longton
Stoke-on-Trent
Staffordshire
ST3 2NA

27 November 2025

Dear Parent / Guardian of Miss Kerry Horner

Miss Kerry Horner, DOB - 25/04/2012, NHS No - 654 109 5612
19 Greendock Street, Longton, Stoke-on-Trent, Staffordshire, ST3 2NA.

Diagnosis: Rubinstein-Taybi Syndrome (RTS) due to CREBBP gene variant

Problems
1. Developmental delay
2. Short stature
3. Microcephaly
4. Distinctive physical features
5. Additional gene variant identified in DDD study of uncertain clincial significance - parental testing ongoing to clarify significance

Surveillance recommended:
1. Dental checkups
2. Hearing surveillance
3. Growth surveillance
4. Developmental surveillance
5. Recommend baseline kidney ultrasound scan - done

Following on from Kerry's genetic clinic consultation, I write to summarise our discussion. This was a review appointment and I met Kerry together with both parents Sarah and Aaron in the clinic at Royal Stoke Hospital. Kerry has a diagnosis of Rubinstein Taybi syndrome (RTS). This was identified

through the Deciphering Developmental Disorders (DDD) research study.
We all have two copies of every gene. We inherit one copy of each gene from our father, and the other copy from out mother. Therefore all our genes come in two copies. We all have two copies of a gene called CREBBP. In Kerry, she has one normal copy of CREBBP but her second copy has a spelling mistake in the genetic code (gene variant). This causes RTS.
Kerry has many physical features in keeping with RTS and therefore this is her underlying diagnosis to explain many of her different issues including her developmental delay, microcephaly (small head circumference), short stature and distinctive physical features.
RTS is a genetic condition that affects many different aspects of an individual. Being short in height is a common feature as well as having developmental delay and learning difficulties. The learning difficulties are can range from moderate to severe. In addition there are physical features that we can see commonly in RTS. These included broad and short thumbs and big toes that can often be angulated. In addition there are facial features that appear commonly in RTS. Looking at Kerry's photos in her genetic file, she does have these facial features and similar thumbs and big toes. Small head circumference (microcephaly) is also usually present. The growth restriction that occurs in RTS usually starts to occur in the first few months of life. Later in childhood or adolescence, the affected children often put on more weight and there is an increased incidence of obesity in RTS.
With regards to surveillance that is required in RTS, it is recommended that Kerry has her development and growth monitored. In addition, hearing checks should be considered periodically as we know that hearing loss can develop in RTS. Regular eye checks should also be considered. Finally, Kerry should have regular check-ups with the dentist as dental problems occurs commonly in RTS.
We discussed that this CREBBP gene change is likely have started for the first time in Kerryrather than inherited. I previously arranged a blood test on both Sarah and Aaron to check this and we are still awaiting the results from this. I will contact you with the results when it is available.
We previously discussed that the DDD study has also identified another gene variant of uncertainclinical significance. This gene can also be associated with short stature, developmental delayand microcephaly but there is not enough evidence to say that this variant is significant (causative) in Kerry. Testing both parents for this other variant will be helpful in clarifying thesignificance of this. If one of you is found to have this other variant then it is less likely to be asignificant finding as neither parent has similar problems as Kerry. I will update you with theresults when available. I suspect that this additional finding is less likely to be significant.
You updated me that Kerry has been started on antibiotics as she has been having recurrent infections. She is due to have a heart scan in August. Her kidney scan was normal which is reassuring. Kerry has a scoliosis (curvature in her spine) and there is plans for an operation as they are concerned the scoliosis may affect her breathing. With regards to her mobility, Kerry is unable to walk very far. She is doing okay at her special needs school. She is unable to write her own name. She wears glasses and previously had a squint that required an operation. Her hearing test was okay and this is being kept under review. She was seen by her dentist for checkups. She is still teething and has the typical talon cusps seen in RTS. Kerry also has constipation and is on Movicol. She is continuing to grow and wears age clothes appropriate for ages 9-10 years. She eats well and has a good appetite. She can get frustrated and therefore this sometimes affects her behaviour.
I appreciate that we covered a lot of information in clinic and hope this letter helps summarise our discussion. There is a support group for RTS in the UK and their website is: rtsuk.org.ukAnother organisation that also has useful information about RTS is called FIND (Further Inform Neurogenetic Disorders) which is based in Birmingham. They have produce a brief fact sheet on RTS (I enclose their one page summary which is quite brief) but further informaiton is available on their website: Findresources.co.uk The specific FIND webpage for RTS information is:
https://www.findresources.co.uk/the-syndromes/rubinstein-taybi-syndrome/key-facts
You may wish to share this letter with Kerry's school to provide more information about RTS.
Please do not hesitate to contact me if you require any further information.

Yours sincerely,

Dr Derek Lim
Consultant Clinical Geneticist

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