International Journal of Science and Research (IJSR)
ISSN: 2319-7064
ResearchGate Impact Factor (2018): 0.28 | SJIF (2019): 7.583

A Study on Rubinstein - Taybi Syndrome

Swapna V Thampi

MSc, M. AM.Ed. (Spl Edu), M.L (LL.M), PGDIHL

Abstract: ***A Study on Rubinstein – Taybi Syndrome Swapna V Thampi* MSc, M. A M.Ed. (Spl Edu), M.L(LL.M), PGDPC, PGDIHL Abstract Rubinstein- Taybi Syndrome also known as Broad Thumb-Hallux syndrome is a genetic disorder that affects many organ systems. It is characterized by facial dysmorphism, beaked nose, slanted eyes, long eye lashes, low hanging nasal septum, extra cusp on lingual side of front tooth, board thumbs and great toes(halluces), and feeding difficulties. In addition to the above there will be growth delays, obesity, intellectual disability, behavioral issues, impulsivity and symptoms of Autism. Further, many persons with RT Syndrome have kidney and heart defects and need medical interventions. Most infants born with this syndrome have a physical appearance which is different from their family at the time of birth. The cause of Rubinstein-Taybi syndrome is due to gene mutation. The medical issues should be addressed as it may evolve in to life threatening complications. Support from occupational therapist, physical therapist and speech therapist along with special education are essential along with parent counselling and training. People with RT Syndrome can achieve autonomy in communication and self-help skills and can be productive with adequate support.***

Keywords: Rubinstein Taybi Syndrome, Genetic disorder, gene mutation, facial dysmorphism, Intellectual Disability

1. Introduction

Rubinstein- Taybi Syndrome also known as Broad Thumb-Hallux syndrome is a genetic disorder affects males and females equally. It is characterized by facial dysmorphism, beaked nose, slanted eyes, long eye lashes, low hanging nasal septum, extra cusp on lingual side of front tooth, board thumbs and great toes(halluces), and feeding difficulties. Affected children have short stature, moderate to severe intellectual disability, distinctive facial features, board thumbs and first toes. In addition to the above there will be obesity in affected children, many children have abnormalities of eyes, dental issues, kidney and heart defects. The cause of Rubinstein-Taybi syndrome is due to gene mutation. One copy of the CREBBP[1] gene is deleted or mutated in persons with Rubinstein-Taybi syndrome. Their cells make only half of the normal amount of CREB binding protein. A considerable amount of reduction in the amount of protein disrupts normal development before and after birth leads to intellectual disability.Other studies establish that mutation in other genes can also cause Rubinstein Taybi syndrome.

Prevalence Rate

The estimated prevalence rate of Rubinstein-Taybi Syndrome is one in 1 lakh to 125,000 births.

History

Rubinstein Taybi Syndrome is named after two medical doctors Jack Rubinstein and Hooshang Taybi[2] who assessed few individuals with certain facial characteristics and broad thumbs and toes along with Intellectual disability and later published research about seven individuals now known asRT syndrome.In the year 1958, Greek Surgeons, Michail, Matsoukas and Theodoru identified persons affected with Rubinstein Taybi Syndrome for the first time in the world

Causes

There are mutations in the CREBBP gene cause about half of cases of this syndrome. The *CREBBP* gene provides instructions for making a protein that helps control the activity of many other genes. This protein, called CREB binding protein, plays an important role in regulating cell growth and division and is essential for normal development before birth. Because one copy of the *CREBBP* gene is deleted or mutated in people with Rubinstein-Taybi syndrome, their cells make only half of the normal amount of CREB binding protein. A reduction in the amount of this protein disrupts normal development before and after birth. Abnormal brain development is thought to underlie intellectual disability in people with Rubinstein Taybi Syndrome. Researchers have not determined how *CREBBP* gene mutations lead to other signs and symptoms.Most cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

2. Diagnosis

When an infant is born with atypical physical appearance and feeding difficulties,parents will approach medical professionals and the infant will be referred for further evaluation. Average age of getting the diagnosis is approximately 15 months. The diagnosis is made after the Genetic testing.

Genetic Involvement

RSTS[3] occurs typically as the result of a de nova pathogenic variant in the family. It is inherited in an autosomal dominant manner. While parents of a child with RSTS are clinically unaffected, siblings are presumed to be at increased risk because of the possibility of a mild phenotype in a heterozygous parent or parent somatic or germline

* Special Education Professional and Lawyer, Chennai

1.The CREBBP gene provides instructions for making CREB binding protein, which regulates the activity of many genes in tissues throughout the body. This protein plays an essential role in controlling cell growth and division and prompting cells to mature and assume specialized functions (differentiate). CREB binding protein appears to be critical for normal development before and after birth. Studies show that this protein is involved in development of the brain and may also be involved in the formation of long-term memories.

[2]Rubinstein JH, Taybi H. Broad thumbs and toes and facial abnormalities. A possible mental retardation syndrome. Am J Dis Child 1963; 105: 588-608

[3] RST- Rubinstein-Taybi syndrome

mosaicism. The empiric recurrence risk for siblings are less than 1%. The risk of offspring is 50%.

Characteristics of RSTS

RSTS is characterized by growth delays, intellectual disability, distinctive facial features like downward slanting eyes, low hanging nasal septum columella[4] , high palate, extra cusp on the lingual side of front tooth - talon cusp[5].Many affected children show hyperactivity, impulsivity, behavioral issues and symptoms of Autism. They may exhibit short attention span, certain self-stimulatory behaviors like rocking, spinning, hand flapping. Many have decreased tolerance of noise and crowds and impulsivity[6].

Management of children with RT Syndrome

The medical issues should be addressed as a priority as it may evolve in to life threatening complications. There need to be Surgeries to repair deformities of toes and fingers in order to relieve pain and improve organ functioning. Help from occupational therapist, physical therapist and speech therapist along with special education is essential. The management includes monitoring feeding and growth, medical intervention which includes treatment eye (annual eye and hearing evaluations), cardiac issues, renal issues, surgical correction of angulated thumbs or duplicated halluces. Many affected children have dysphagia- feeding difficulties. Many of them show short span of attention. Behavioral issues need to be addressed. As children with RT Syndrome have intellectual disability, they need remedial therapeutic intervention till achieve autonomy in life. Further, early intervention program, special education,along with parent counselling and parent training is required.

3. Conclusion

RT Syndrome is a rare genetic disorder due to gene mutation. Most infants born with this syndrome have a physical appearance which is different from their family at the time of birth. This condition is considered to have an autosomal dominant pattern of inheritance. There is not muchInformation regarding adults with this syndrome is available. There is a need to create more Awareness about this syndrome. If the medical problems are treated and adequate care and special education is provided with positive family support most affected individuals can achievemaximum independence in self-help skills and social communication areas.Many children with RT Syndrome can perform well in supervised settings. Due to technological advancement parents can access more information about the condition and can be in touch with other parents and professionals. Support and advocacy groups of families with RT Syndrome is highly essential. There should be a platform for parents to interact and share their experiences in raising a child with this syndrome.

References

[1] Rubinstein JH, Taybi H(1963).Broad thumbs and toes and facial abnormalities. American Journal of Diseases of Children 105:588-608
[2] Hennekam RC, Lommen EJ, Strengers JL, Van Spiker HG, Jansen Kokx TM. Rubinstein Taybi Syndrome in a mother and son. European Journal of Pediatrics 1989; 148:439-41
[3] Hsiung SH. Rubinstein Taybi Syndrome Dermatol online Journal 2004;10:2
[4] Sharma N, Mali AM, Bapat SA, Spectrum of CREBBP Mutations in Indian Patients with Rubinstein Taybi Syndrome. J Biosci2010;35:187-202
[5] https://rarediseases.org/rubinstein-taybi-syndrome

[4]nasal septum columella-low hanging nasal septum

5. Talon cusp is an uncommon developmental dental abnormality, showing a cusp-like accessory structure, varying in size from a prominent cingulum to a marked projection affecting the lingual surface of the maxillary and mandibular teeth

[6]https://rarediseases.org/rubinstein-taybi-syndrome