Rubenstein-Taybi Syndrome
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Introduction:
Rubinstein-Taybi syndrome (RTS) is a rare congenital disorder involving a combination of both intellectual impairment and physical abnormalities. It is caused by a microdeletion in the gene encoding for CREB Binding Protein (CREBBP) found on chromosome 16p13.3. Typical features include broad, angulated thumbs and halluces, short stature, beaked nose, small maxilla, talon cusps, and down-slanting palpebral fissures. In addition, many individuals with RTS have a propensity toward keloid formation.

Case Report:
In this case we present a 38-year-old Caucasian male with distinct physical characteristics who presented to our clinic to have his keloids treated. His past medical history is varied but is typical of the syndrome. He was born without tear ducts. He was born with hard palate and jaw deformities, with undescended testicles and without an Odontoid process. Seven years prior the patient was noted to have severe obstructive sleep apnea secondary to a significantly enlarged lingual tonsil. Due to the size of the tonsil, during the surgery, the patient's epiglottis was damaged and scarred open. A feeding tube had to be placed to prevent aspiration. He is currently undergoing chemotherapy for stage IV non-Hodgkin’s lymphoma (NHL).

Family history: Cousin diagnosed with Ehlers-Danlos syndrome; all else unremarkable
Social history: graduated from high school; attends church; very sociable and has a great memory
Medications: Rituax Q2 months for NHL; testosterone crème

Differential Diagnosis:
Conditions with Broad thumbs:
- Rothmund-Thomson syndrome
- Poikiloderma Congenital
- Stapes ankylosis with broad thumb and toes

Conditions with Short Stature:
- Bloom Syndrome
- Trichothiodystrophy (PIBIDS)
- Cockayne Syndrome

Distinct Facial Features:
- Down slanted palpebral fissures
- Low hanging columella
- High palate
- Grimacing smile

RTS is an inherited disorder in either a sporadic or autosomal-dominant fashion resulting in a CREBBP defect. RTS is characterized by distinct facial features, broad angulated thumbs and great toes, short stature, intellectual impairment, keloid formation, congenital heart defects, vascular malformations, cryptorchidism, and increased susceptibility to solid tumors. Birth prevalence is uncommon occurring in 1 in 100,000 to 125,000 births.

“CREBBP (CRE binding protein or CBP) encodes for a large ubiquitously expressed protein of the same name that performs multiple roles in transcriptional co-activation, including the acetylation of histone and nonhistone targets… Somatic mutations in CREBBP have recently been noted in more than one third of diffuse large-cell non-Hodgkin lymphoma and follicular lymphoma at diagnosis.”

Conclusion
RTS is an uncommon congenital disorder with no known cure characterized by intellectual impairment and multiple physical deformities. It has a number of dermatologic manifestations including keloid formation. The patient presented in this case had several keloids on his abdomen that were in the process of being treated with intralesional steroid injections to which he appeared to be responding well.

References