Chapter 1 : Axenfeld, Israel | racedaydvl.com

*axenfeld, israel He was born in Nemirov and was originally a follower of the á¸¥asidic rabbi *Naá¸¥man of Bratslav, in Podolia, Ukraine, but after traveling through Germany as a supplier of the Russian Army in and coming into contact with the early maskilim of Brody, Galicia, Axenfeld became staunchly anti-á¸¥asidic.*

Genes and Databases for chromosome locus and protein. See Molecular Genetics for information on allelic variants detected in this gene. Sequence analysis detects variants that are benign, likely benign, of uncertain significance , likely pathogenic, or pathogenic. For issues to consider in interpretation of sequence analysis results, click here. Included in the variety of methods that may be used are: Thus, the current limited understanding regarding the spectrum of the phenotype and its natural history would be expected to evolve over time as more affected individuals are reported in the medical literature. Intrauterine growth restriction IUGR. Mild-moderate short stature is usually present throughout childhood. Bone age may or may not be delayed. Mild short stature has been seen in most adults reported to date. Adult height in males with a molecularly confirmed diagnosis of SHORT syndrome was between and cm and in females between and cm [Chudasama et al , Dyment et al , Thauvin-Robinet et al]. Lack of subcutaneous fat is evident in the face and later becomes more readily apparent in the chest and upper extremities. Although the buttocks and legs are often spared, localized regions of lipoatrophy at the elbows and buttocks have been reported [Aarskog et al , Koenig et al]. The hands also lack subcutaneous fat, and the skin has an aged, translucent appearance. The body habitus is described as thin. All four adult males reported to date with a molecularly confirmed diagnosis had a body mass index BMI below The characteristic facial features of SHORT syndrome â€” sometimes described as having an "aged" or "progeroid" appearance [Koenig et al] â€” are present at birth and become increasingly apparent with age. Head shape is normal and occipital-frontal circumference is proportionate with other growth parameters. Although insulin resistance may be evident in mid-childhood or adolescence, diabetes mellitus typically does not develop until early adulthood [Aarskog et al , Schwingshandl et al]. Axenfeld-Rieger anomaly or related anterior chamber ocular anomalies, also referred to as anterior chamber dysgenesis, have been reported in the majority of individuals with SHORT syndrome. Glaucoma, which has been reported in at least one individual at birth [Brodsky et al], can also develop later [Bankier et al]. Glaucoma is thought to be the result of poorly developed aqueous humor drainage structures of the anterior chamber of the eye. To date, delayed dentition has been observed in all individuals with a molecularly confirmed diagnosis of SHORT syndrome. Other dental issues include hypodontia, enamel hypoplasia, and malocclusion. Multiple dental caries have also been reported [Koenig et al]. Hearing loss of dB has been diagnosed within the first year of life. Among individuals with a molecularly confirmed diagnosis of SHORT syndrome, two have hearing loss; both have the recurrent c. Although there does not appear to be an increased risk for life-threatening infections or evidence of clinical immunodeficiency, there have been reports of a nonspecific history of frequent infections [Koenig et al]. Nephrocalcinosis was identified incidentally in the son at age two months on an abdominal US examination performed for follow up of anorectal atresia ; the nephrocalcinosis was stable when reassessed at age two years. The mother also developed nephrocalcinosis as an adult. Pulmonic stenosis and ectopic kidney have also been reported [Koenig et al , Schroeder et al]. The recurrent missense pathogenic variant , c. While individuals with this specific variant usually have typical SHORT syndrome, to date the numbers are too small to determine whether the phenotype observed with this pathogenic variant differs from that observed with other pathogenic variants. Penetrance The penetrance of SHORT syndrome appears complete in all individuals undergoing molecular genetic testing to date: All simplex cases i. Nomenclature Since it was first described in the early s, what appears to be SHORT syndrome has been described by different terms including: No ethnic predilection is known. Genetically Related Allelic Disorders To date, two individuals with different phenotypes have been reported with germline pathogenic variants in PIK3R1. One individual with an amino acid substitution in the N-terminal SH2 domain of PIK3R1 had acanthosis nigricans and severe insulin resistance [Baynes et al]. Differential Diagnosis Russell-Silver syndrome. Intrauterine growth restriction IUGR and postnatal growth deficiency are frequent features of

Russell-Silver syndrome. Individuals with Russell-Silver can also exhibit overlapping nonspecific facial features including a triangular-shaped face. Since it is possible that some individuals with a clinical diagnosis of Russell-Silver and normal molecular studies have a diagnosis of SHORT syndrome, careful consideration of the facial phenotype in these patients is warranted. Alagille syndrome is a complex multisystem disorder involving the liver, heart, eyes, face, and skeleton. It is inherited in an autosomal dominant manner. Anterior chamber eye anomalies. Both conditions are inherited in an autosomal dominant manner. Copy number variations at several loci including PITX2 4q25 and BMP4 14q22 which are frequently detected by chromosomal microarray analysis can lead to syndromic anterior chamber eye anomalies with phenotypes that to date have been clinically distinct from those of SHORT syndrome [Karadeniz et al , Lines et al , Reis et al]. Berardinelli-Seip congenital lipodystrophy is usually diagnosed at birth or shortly thereafter and is characterized by severe generalized lipodystrophy, hepatomegaly, and acromegaloid facial features. As in SHORT syndrome, insulin resistance and subsequent diabetes mellitus become common in late adolescence and early adulthood. Typically, this syndrome results from a heterozygous de novo pathogenic variant in LMNA. Clinical features develop in childhood and resemble some features of accelerated aging with disease progression that is distinct from that of SHORT syndrome. In contrast, individuals with SHORT syndrome typically have phenotypic features evident at birth including the lipodystrophy and underdeveloped alae nasae. The facial features of SHORT syndrome can be confused with those seen in Hutchinson-Gilford progeria as well as other syndromes including Johansson-Blizzard syndrome caused by biallelic pathogenic variants in UBR1 and Hallerman-Streiff syndrome for which no genetic cause has been identified. Management Evaluations Following Initial Diagnosis To establish the extent of disease and needs in an individual diagnosed with SHORT syndrome the following evaluations are recommended if not performed at the time of diagnosis: Measurement of length and weight Assessment of speech and language or motor skills in those with evidence of developmental delays For older children and adults just diagnosed with SHORT syndrome: Treatment by an ophthalmologist experienced in management of developmental eye disorders or glaucoma is indicated to reduce and stabilize ocular pressures and to preserve vision. Treatment of dental anomalies is standard and may include crowns and dental prostheses. Standard treatment for glucose intolerance and diabetes mellitus with diet, lifestyle, oral medication, and insulin under the supervision of a specialist in diabetes care is recommended. See Hereditary Hearing Loss and Deafness. Surveillance The following are appropriate: Evaluation of Relatives at Risk See Genetic Counseling for issues related to testing of at-risk relatives for genetic counseling purposes. If present, diabetes mellitus is managed as appropriate. There may not be clinical trials for this disorder. Genetic Counseling Genetic counseling is the process of providing individuals and families with information on the nature, inheritance, and implications of genetic disorders to help them make informed medical and personal decisions. The following section deals with genetic risk assessment and the use of family history and genetic testing to clarify genetic status for family members. This section is not meant to address all personal, cultural, or ethical issues that individuals may face or to substitute for consultation with a genetics professional. If the pathogenic variant found in the proband cannot be detected in leukocyte DNA of either parent, two possible explanations are germline mosaicism in a parent or a de novo pathogenic variant in the proband. Although no instances of confirmed germline mosaicism have been reported, it remains a possibility. Of note, the occurrence of SHORT syndrome in a sib pair reported in by Gorlin et al [] appears to have been due to germline mosaicism. The proportion of cases caused by de novo pathogenic variants is unknown. Evaluation of parents may determine that one is affected but has escaped previous diagnosis because of a milder phenotypic presentation. Therefore, an apparently negative family history cannot be confirmed until appropriate evaluations have been performed. To date the clinical status of all affected individuals has been correctly ascertained on the basis of physical examination primarily presence of short stature, lipodystrophy, and characteristic facial features. When the parents are clinically unaffected, the risk to the sibs of a proband appears to be low. Recurrence in a sib in the absence of an affected parent has not been observed to date. The sibs of a proband with clinically unaffected parents are still at increased risk for SHORT syndrome because of the theoretic possibility of reduced penetrance in a parent. If the pathogenic variant found in the proband cannot be detected in the leukocyte DNA of either parent, the risk to sibs is low

but greater than that of the general population because of the possibility of germline mosaicism. Offspring of a proband. Related Genetic Counseling Issues Considerations in families with an apparent de novo pathogenic variant. When neither parent of a proband with an autosomal dominant condition has the pathogenic variant or clinical evidence of the disorder, the pathogenic variant is likely de novo. However, possible non-medical explanations including alternate paternity or maternity e. Family planning The optimal time for determination of genetic risk and discussion of the availability of prenatal testing is before pregnancy. It is appropriate to offer genetic counseling including discussion of potential risks to offspring and reproductive options to young adults who are affected or at risk. DNA banking is the storage of DNA typically extracted from white blood cells for possible future use. Because it is likely that testing methodology and our understanding of genes, allelic variants, and diseases will improve in the future, consideration should be given to banking DNA of affected individuals. Prenatal Testing and Preimplantation Genetic Diagnosis Once the PIK3R1 pathogenic variant has been identified in an affected family member, prenatal testing for a pregnancy at increased risk and preimplantation genetic diagnosis are possible. Differences in perspective may exist among medical professionals and within families regarding the use of prenatal testing, particularly if the testing is being considered for the purpose of pregnancy termination rather than early diagnosis. While most centers would consider decisions regarding prenatal testing to be the choice of the parents, discussion of these issues is appropriate. GeneReviews is not responsible for the information provided by other organizations. For information on selection criteria, click here.

Chapter 2 : I, an Israeli citizen, joined ISIS and lived to tell the tale

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A talented scientist and a private individual, Resnik gave her deepest loyalties to her father, her career, and her close friends. Although she avoided publicity whenever possible, she was fun-loving and spirited; her romantic crush on actor Tom Selleck was often a source of good-natured teasing among her coworkers. Born Judith Arlene Resnik on April 5, , in Akron, Ohio, to first-generation Jewish Russian parents, Judith was a bright, curious child who, by kindergarten, could both read and solve simple math problems. Her father, Marvin, was an optometrist and a part-time cantor when he married Sarah Polensky, a former legal secretary from Cleveland Heights. After Judith was born, the Resniks had a son, Charles. The Resniks were an upper-middle-class Jewish family devoted to their religion and to all learning. Gifted in math and science, Resnik excelled in academics from a young age. She also attended Hebrew school and, by her teenage years, was an accomplished classical pianist. Teachers and friends described her as extremely bright, disciplined, perfectionistic, and personable. After graduating in , she married Michael Oldak, a fellow engineering student. In , they moved to Washington, D. She and Oldak divorced in In , while Resnik was finishing work on her doctorate, NASA began recruiting minorities and women to the space program. Though she had never shown particular interest in the space program, she decided to apply. After receiving academic honors for her doctoral work in electrical engineering, she accepted a job with Xerox. In , at age twenty-nine, Resnik was one of six women accepted into the program. Resnik would be the second American woman to fly in space after Sally Ride in , and the fourth woman worldwide. During her first six years at NASA, she specialized in the operation of a remote-control mechanical arm that moved objects located outside the spacecraft. Famous for its civilian crew member, teacher Christa McAuliffe, the mission endured three delays before taking off at Seventy-three seconds into the flight, the space shuttle exploded in midair due to hydrogen leakage caused by faulty O-ring seals. Along with her six crew members, Resnik died in one of the worst space disasters in history. Bibliography Bernstein, Joanne E. Judith Resnik, Challenger Astronaut Telephone interview by author, June 27, Spencer, Scott, and Chris Spolar. Science , Engineering More on Judith Resnik.

DOWNLOAD PDF AXENFELD, I. AND REZNIK, L. RECRUITS.

Chapter 3 : Prediction of elastic properties of pyrolytic carbon based on orientation angle - IOPscience

Axenfeld-Rieger syndrome is a genetically heterogeneous, autosomal dominant disorder characterized by anomalies of the anterior segment of the eye, face, teeth, and umbilicus.

Published online Aug Venugopalan ,1 Huojun Cao ,1 Flavia O. Pinho ,1 Michael L. Paine ,2 Malcolm L. Snead ,2 Elena V. Semina ,3 and Brad A. Semina Find articles by Elena V. Published by Oxford University Press. For Permissions, please email: ARS is genetically associated with mutations in the PITX2 gene, which encodes one of the earliest transcription factors to initiate tooth development. Thus, Pitx2 has long been considered as an upstream regulator of the transcriptional hierarchy in early tooth development. However, because Pitx2 is also a major regulator of later stages of tooth development, especially during amelogenesis, it is unclear how mutant forms cause ARS dental anomalies. In this report, we outline the transcriptional mechanism that is defective in ARS. We demonstrate that during normal tooth development Pitx2 activates Amelogenin Amel expression, whose product is required for enamel formation, and that this regulation is perturbed by missense PITX2 mutations found in ARS patients. We further show that Pitx2-mediated Amel activation is controlled by chromatin-associated factor Hmgn2, and that Hmgn2 prevents Pitx2 from efficiently binding to and activating the Amel promoter. Consistent with a physiological significance to this interaction, we show that KHmgn2 transgenic mice display a severe loss of Amel expression on the labial side of the lower incisors, as well as enamel hypoplasiaâ€”consistent with the human ARS phenotype. Collectively, these findings define transcriptional mechanisms involved in normal tooth development and shed light on the molecular underpinnings of the enamel defect observed in ARS patients who carry PITX2 mutations. Moreover, our findings validate the etiology of the enamel defect in a novel mouse model of ARS. Patients with ARS exhibit a wide spectrum of developmental defects 3 , 4 , including umbilical anomalies, ocular defects and craniofacial abnormalities 5 , 6. Classic clinical presentations also include various dental defects, often involving hypoplasia of the enamel 6 , 7. Intriguingly, most of the dental phenotypes associated with ARS include enamel defects that are clinically similar to those observed in patients with Amelogenesis Imperfecta AI 8. PITX2 is a bicoid-motif-binding protein and a member of the paired-like homeobox transcription factor family It is also one of the earliest epithelial markers of tooth development 20 , and plays fundamental roles in the genetic control of pattern formation and epithelial differentiation in the tooth 19 , 21 , Given that Pitx2 has long been considered as a master regulator of the transcriptional hierarchy in early tooth development 23 , 24 , PITX2 mutations are thought to account for many of the tooth defects in ARS patients. Pitx2 is highly expressed in the cervical loop region of the developing tooth epithelium a stem cell niche and initiates differentiation. Once the epithelial stem cells start to migrate toward the apical tip of the tooth, Pitx2 expression decreases and is maintained at a relatively low level. Nevertheless, we found that Pitx2 is a major determinant of the later stages of tooth development, especially during amelogenesis enamel formation. ARS patients with these mutations have classical ARS manifestations, including a variety of dental defects: During tooth organogenesis, a spectrum of proteins other than Pitx2, including signaling molecules and additional transcription factors, contribute to the regulation of amelogenesis 28 â€” Dlx2 and FoxJ1 are both transcriptionally regulated by Pitx2, and their protein products interact physically with each other and with Pitx2, forming a complex in which hierarchical interactions take place to regulate amelogenesis 34 , Wnt signaling is also required, upstream of these events and specifically for the late stages of tooth development 36 , Tooth development depends on the activities of not only transcription factors, but also chromatin-remodeling proteins One candidate for such activity is Hmgn2, a chromatin-associated high-mobility group protein that binds to histones 41 , Many transcription factors of the homeodomain family bind to Hmgn2 with high affinity, and form inactive complexes with histones on the open chromatin Hmgn2 inhibits homeodomain transcription factors from binding DNA, but recruits transcription factors to chromatin where they are poised to regulate transcription upon interaction with other factors that de-repress the Hmgn2 inactive complex to allow the transcription factors to bind DNA. Hmgn2 is highly expressed in the dental epithelium during early embryogenesis, but its levels decrease similar to Pitx2 as development proceeds. In

the current study, we investigated the functional significance of the Pitx2 transcriptional hierarchy with respect to tooth development, including the roles that the Pitx2 co-factors play in regulating amelogenesis. Although the focus on the molecular-genetic aspects of ARS has increased in recent years, the molecular underpinnings of these particular tooth defects in ARS remain largely unknown. Hmgn2 alters Amel expression by interacting with Pitx2 during amelogenesis, and that the overexpression of Hmgn2 in vivo could recapitulate the effects of Pitx2 loss-of-function, with such animals serving as a model for further studies of ARS dental defects. Ideally, a Pitx2 loss-of-function mouse would have served as an in vivo model for the study of such defects.

Chapter 4 : Ithaca College Athletics

Morgan Axenfeld's high school sports timeline. MaxPreps has events and updates about Morgan Axenfeld while she was playing lacrosse and basketball at Fayetteville-Manlius High School dating as far back as

Oncotarget Abe, T. Biomaterials Ackermann, S. Cereb Cortex bhu Adeyemi, R. J Cell Physiol Ahmad, G. Alam, Majid Botchkareva, Natalia V. Journal of Cell Science Akasaki, Y. Bardeesy, Nabeel Bronson, Roderick T. Liu, Wei Losman, Julie A. Zhang, Haikuo Dhydroxyglutarate produced by mutant IDH2 causes cardiomyopathy and neurodegeneration in mice. Nat Protoc Akil, O. Stable tagged ubiquitin exchange system for the global investigation of cellular ubiquitination. Kitade, Mitsuteru Marquardt, Jens U. Seo, Daekwan Thorgeirsson, Snorri S. Peters, Christoph Quick, Marsha L. Wang, Hao-Wei Distinct functions of macrophage-derived and cancer cell-derived cathepsin Z combine to promote tumor malignancy via interactions with the extracellular matrix. Eukaryotic Cell Alberti, L. Cell Rep Alebrahim, S. Metcalfe, Su Philpott, Anna The phosphorylation status of Ascl1 is a key determinant of neuronal differentiation and maturation in vivo and in vitro. Development Allen, S. Carcinogenesis bgu Althubiti, M. Cancer Research Alzhanova, D. Fallahi, Mohammad Kaye, Frederic J. Wu, Lizi Young, Brandon M. Nat Commun Applebaum, M. Chen, Gang Cramer, Samuel W. Wang, Xinming Zink, Anastasia N. Zu, Tao Mutant? The Journal of Neuroscience Arnoldo, A. Genome Med 32 Asokan, S. Dev Cell Aspesi, A. Cancer Atmadibrata, B. Stem Cell Reports Auslander, D. Nat Methods Axelrod, M. Cancer Cell Baboo, S. J Orthop Res Bailey, R. Mol Neurodegener 8 Arteaga, Carlos L. Cronin, Maureen Doimi, Franco D. Kurupi, Richard Lehmann, Brian D. Owens, Phillip Palmer, Gary A. Proteomics Bachmann, A. The Journal of Neuroscience Apostolou, E. Stem Cell Reports Beeharry, Y. Virology Behrens, A. Stem Cells Dev Advani, V. Nature Benaich, N. Cell Rep Benevento, M. Itzhak, Inbal Jung, Jae U. Stem Cells Akbarian, S. Ann Rheum Dis annrheumdis Ambrogio, C. Cell Rep Bennion, B. Respir Res Bojovic, B. Cell Rep Bonardi, D. Das, Pritam Golde, Todd E. Guo, Qinxi Jankowsky, Joanna L. The Journal of Neuroscience Amit, I. Mol Cell Bosnakovski, D. Skelet Muscle 4 Bosson, A. Mol Cell Atadja, P. Blood Bernard, St? Journal of Biological Chemistry Bowles, J. Dev Biol Boyd, Justin D. Ebata, Atsushi Feiler, Marisa S. Kraemer, Brian Lee-Armandt, J. Gonia, Sara Gow, Neil A. Li, Bilan Reynolds, Susan D. Cell Metabolism Brown, J. Journal of Biological Chemistry Azam, S. Behav Brain Res Bruch, J. Journal of Virology Brundage, M. Cell Stem Cell Abraham, B. Cell Stem Cell Bindokas, V. Mol Neurodegener 1 Ayaz, B. Cell Death Dis e Bunjobpol, W. Cell Death Differ Burugula, B. Mol Cell Biol Busskamp, V. Van de Casteele, M. Diabetologia Callegari, S. Clinical Cancer Research Andrews, S. Journal of Virology Bell, Andrew I. Hakimjavadi, Roya Loughran, Sin? Phelan, Susan Smith, Sin? Journal of Virology Canalis, E. J Cell Physiol Canalis, E. Journal of Virology Carey, T. Stem Cells Dev Carver, J. Circulation Research Castellana, D. Neuron Cebrian, J. Cekanaviciute, Egle Dietrich, Hans K. Egusquiza, Riann Koshy, Anita A. The Journal of Immunology Amelio, I. J Neurophysiol Boscolo, Francesca S. Xiang, Wanyi Early forming label-retaining muscle stem cells require p27kip1 for maintenance of the primitive state. Development Chakraborty, S. Jailwala, Parthav Kennedy, Mark W. Si, Han Yamaguchi, Terry P. Development Arany, Z. Skelet Muscle 2 Chan, David C. Hess, Sonja Sweredoski, Michael J. Journal of Biological Chemistry Ahlenius, H.

Chapter 5 : Los Angeles Times - We are currently unavailable in your region

Axenfeld-Rieger syndrome is a term that can be used to describe a variety of overlapping phenotypes. To date, at least three known genetic loci can cause these disorders.

Chapter 6 : A model for the molecular underpinnings of tooth defects in Axenfeldâ€“Rieger syndrome

I, an Israeli citizen, joined ISIS and lived to tell the tale. An Israeli recounts slipping into Syria under fire, fighting his first battle, taking part in a beheading and making an unlikely return home.

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Chapter 7 : William & Mary Athletics - William and Mary Tribe Women's Soccer All-Time Roster

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SHORT syndrome is a mnemonic for short stature, hyperextensibility, ocular depression (deeply set eyes), Rieger anomaly, and teething delay. Axenfeld-Rieger.

Chapter 9 : SHORT Syndrome - GeneReviewsÂ® - NCBI Bookshelf

â€¢ Managed the presentation, selection, offer, negotiation, closing, and administrative components involved in full lifecycle recruiting of health care personnel (RN, NP, LPN, MSW).