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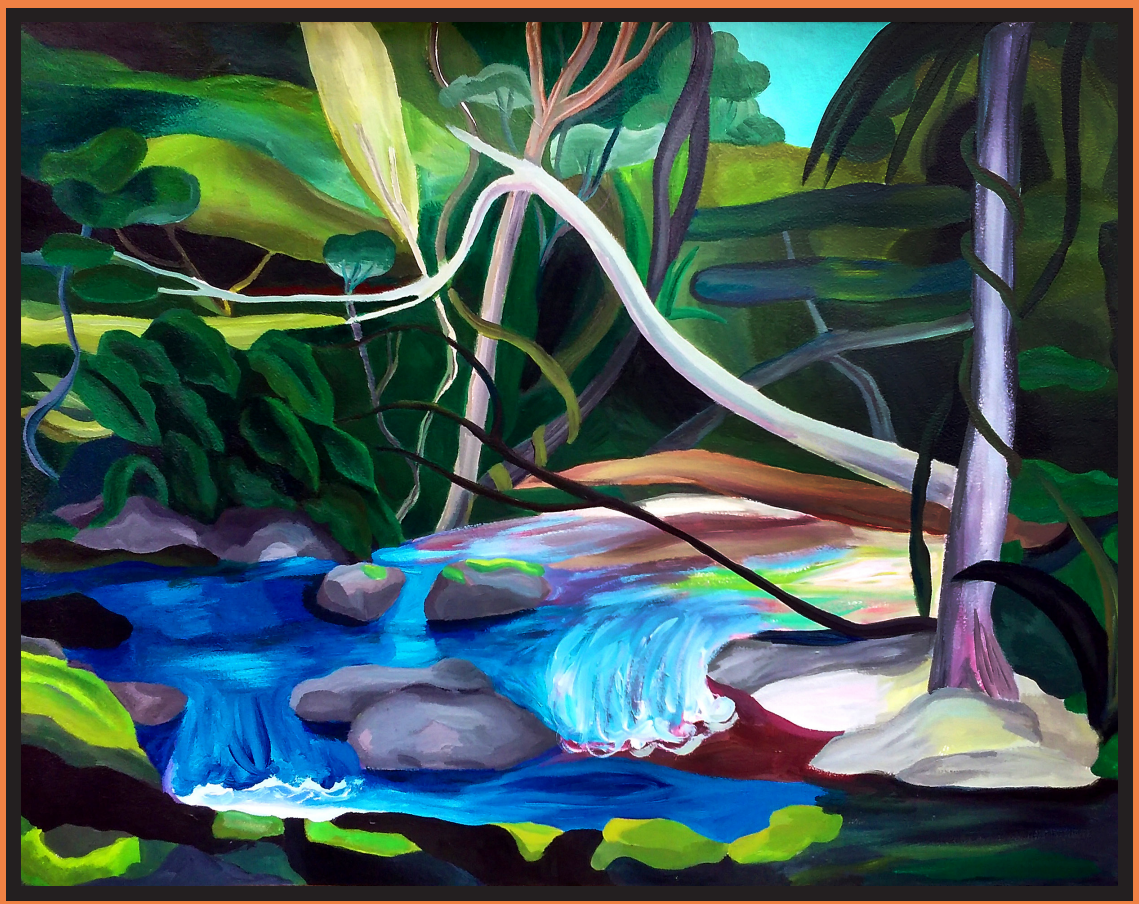

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REUNIÓN CONJUNTA SAIC SAB AAFE AACYTAL 2023

LXVIII REUNIÓN ANUAL DE LA
SOCIEDAD ARGENTINA DE INVESTIGACIÓN CLÍNICA
(SAIC)

XXV JORNADAS ANUALES DE LA SOCIEDAD
ARGENTINA DE BIOLOGÍA
(SAB)

LV REUNIÓN ANUAL DE LA ASOCIACIÓN
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VIII REUNIÓN CIENTÍFICA REGIONAL DE LA
ASOCIACIÓN ARGENTINA DE CIENCIA Y
TECNOLOGÍA DE ANIMALES DE LABORATORIO
(AACYTAL)

15-17 de noviembre de 2023
Hotel 13 de Julio – Mar del Plata

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not been reached after deep phenotyping. Furthermore, reverse phenotyping allowed the identification of a clinically inapparent congenital heart defect in one case.

COMPREHENSIVE APPROACH FOR THE GENETIC DIAGNOSIS OF PATIENTS WITH WAARDENBURG SYNDROME

Paula Buonfiglio[1], Agustín Izquierdo[2,3], Mariela Pace[1], Vanesa Lotersztein[4], Paloma Brun[5], Ana Belén Elgoyhen[1,6], Viviana Dalamón[1]

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Waardenburg syndrome (WS) is one of the most common syndromic forms of genetic hearing loss (HL), accounting nearly for 2-5% of congenital HL. It is characterized by the presence of hearing impairment in association with pigmentation abnormalities that may affect the skin, hair, and/or eyes. WS is divided into four subtypes according to different concomitant phenotypes and its generally of autosomal dominant inheritance. Up to date, seven genes are related to WS: *PAX*3*, MITF, EDNRB, ENDR, SOX*10*, KITLG and SNAI2.* Disease-causing variants are mainly single nucleotide variants (SNVs), though copy number variants (CNVs) have also been reported. The aim of this work is to identify the genetic causes of WS in four family cases with a dominant mode of inheritance.
As the first step Whole Exome Sequencing (WES) was performed for SNVs screening, filtering out the target genes. When negative, CNVs were analyzed using DECoN tool on WES raw data. Multiplex ligation-dependent probe amplification (MLPA) was carried out to confirm and segregate CNVs identified in the family members.
Three of the 4 families analyzed carried heterozygous pathogenic variants: one SNV and two CNVs in the WS target genes. In family #1 a stop variant (NM_001354604.2:c.1198C>T p.Arg400*) was detected in *MITF* and segregated in one affected son of the family. In family #2 a deletion of 1 exon in *PAX*3 gene was detected and segregated also in the affected mother. In family #3, remarkably, a large novel deletion comprising 7 genes including *SOX*10 was detected in the exome CNVs analysis. The complete loss of *SOX*10 was confirmed and also segregated in the affected family members by MLPA. The combination of techniques and bioinformatic analysis resulted in a better diagnostic rate and in a substantial improvement in the molecular diagnosis of patients. These results highlight the importance of combining different strategies to achieve diagnosis leading to an accurate genetic counseling.

SAIC AWARD - Fundación BIGAND - Multidisciplinary call for young investigators.
Friday 17th November 16:00-18:30
Juries: Rodolfo Rey, Marta Tesone, Edith Kordon

PROTEIN KINASE D1 ACTIVITY IS ASSOCIATED TO MOUSE SPERM CAPACITATION

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Sperm are specialized and transcriptionally inactive cells dependent on post-translational modifications, such as protein phosphorylation, to carry out their functions. These changes occur during the process called sperm capacitation. The protein kinase D family (PKDs) includes three serine/threonine kinases (PKD1, PKD2 and PKD3) being PKD1 the most studied and ubiquitous one. This family of kinases are involved in fundamental biological process including signal transduction, cytoskeleton remodeling, golgi transport and oxidative stress among other functions. Recent results from our laboratory indicate that PKD1 is present in the sperm of human, mice, equine and bovine. Since there are no previous reports about the presence and function(s) of any PKD in mammal sperm, we examined its distribution and function(s).
We observed that: 1) PKD1 is present and catalytically active (p-PKD) in bovine, equine, mice, and human sperm (IIF/WB). 2) The capacitation process induces changes in kinase localization and activation levels, evaluated by super resolution microscopy in mouse sperm. 3) Mouse sperm incubated in the presence of specific PKD inhibitors (Kb142-70 and CRT0066101) before spermatozoa ca-