Nro panel	ID Trabajo Genética	Título	Presentador	Mención
	P1	Posters		
	Fecha y hora:	**19/11/2024 11:00-12:00 h**		
	Coordinadores:	**Cecilia Fernández, Carlos David Bruque, Paula Buonfiglio**		
97	130	**ZEBRAFISH AS AN IN VIVO MODEL TO CHARACTERIZE THE PATHOGENICITY OF IGF1 VARIANTS IDENTIFIED IN PATIENTS WITH SEVERE SHORT STATURE**	Macarena Recalcatti (Centro de Inve	Si
99	344	**HEALTH-RELATED QUALITY OF LIFE IN TRANSTHYRETIN AMYLOIDOSIS: PATIENT-REPORTED OUTCOMES FROM THE REGISTRY OF PEOPLE WITH AMYLOIDOSIS**	Marcelina Carretero (Hospital Italian	No
101	240	**Molecular characterization of the breakpoints in F9 deletions in three unrelated families with haemophilia B**	Betiana Michelle Ziegler (Instituto de	No
103	445	**CHROMATINOPATHIES: PATHOGENIC COPY NUMBER VARIANTS IN GENES ASSOCIATED WITH CHROMATIN REMODELING**	Barbara Casali (Centro de Investiga	No
105	465	**MOLECULAR AND CLINICAL INSIGHTS INTO LAMA2-RELATED DYSTROPHIES IN AN ARGENTINEAN PAEDIATRIC COHORT**	María Eugenia Foncuberta (Laborat	No
107	371	**BEYOND THE BASICS: EXPLORING STRC-RELATED HEARING LOSS, A LESSER-KNOWN CAUSE OF HEARING LOSS IN ARGENTINA.**	Sofia Grinberg (Laboratorio de Fisio	No
109	71	**Frequency and characteristics of double IGHV rearrangements in chronic lymphocytic leukemia patients.**	Carmen Stanganelli (División Patolo	No
	P2	Posters		
	Fecha y hora:	**20/11/2024 11:30-12:30 h**		
	Coordinadores:	**Viviana Dalamon, Florencia Giliberto**		
97	452	**CELLULAR CHARACTERIZATION OF THE EXON13_15DUP OF LDLR ASSOCIATED WITH FAMILIAL HYPERCHOLESTEROLEMIA.**	Carolina Andrea Alarcón Reyes (Uni	No
99	162	**Design and application of a new protocol for the detection of variants by Next Generation Sequencing (NGS) in patients with Hemophilia A (HA) and B (HB).**	Liliana Carmen Rossetti (Instituto de	No
101	217	**EPIGENETIC INSIGHTS INTO SPORADIC COLORECTAL CANCER USING EPICV2 BEADCHIP: A FIRST IN-LINE ANALYSIS OF AN EIGHT-GENE CIMP-LINKED PANEL**	Walter Hernán Pavicic (Instituto de M	No
103	232	**CONTRIBUTION OF LARGE REARRANGEMENTS IN BRCA1/2 GENES AND CHEK2 1100DELC ALLELE VARIANT TO THE DEVELOPMENT OF BREAST/OVARIAN CANCER IN UNSELECTED ARGENTINIAN POPULATION**	Luciana Berlanga (Centro Nacional I	No
105	16	**Risk of Sudden death: Cohort Analysis of Genetic and Clinical Outcomes**	Luis Enrique Gomez (1. Unidad de C	No
	P3	Posters		
	Fecha y hora:	**20/11/2024 16:10-17:10 h**		
	Coordinadores:	**Fiorella Belforte, Lourdes Correa Brito**		
97	440	**DISCOVERING A NOVEL VARIANT IN HEREDITARY COPROPORPHYRIA: DIAGNOSTIC INSIGHTS**	LAURA VARELA (1Centro de Invest	No
99	372	**"UNDERSTANDING THE COMPLEXITIES OF HEARING LOSS DIAGNOSIS WITH WHOLE EXOME SEQUENCING: SYNDROMIC OR NOT?"**	Mariela Pace (Laboratorio de Fisiolo	No
101	532	**GENETIC AND BIOCHEMICAL CHARACTERIZATION OF ARGENTINE PATIENTS WITH GLYCOGEN STORAGE DISEASES TYPE VI AND IX**	Silene Maite Silvera Ruiz (Centro de	No
103	513	**FROM POLICY TO PRACTICE: ADVANCEMENTS IN THE MANAGEMENT OF RARE DISEASES.**	Sabrina Soledad Fernández (Unidad	No
105	520	**INTEGRATIVE EVALUATION OF GUT METAGENOMIC BIOMARKERS IN ULCERATIVE COLITIS ASSOCIATED WITH SECRETORY IGA GLYCOSYLATION IN THE CONTEXT OF TRANSKINGDOM COMMUNICATION NETWORKS.**	Valeria Cristina Soler Rivero (GeC-U	No
107	391	**CHALLENGES IN THE DESIGN OF A TARGET microRNA AND GENE EXPRESSION SYSTEM IN BACULOVIRAL VECTORS TO INDUCE PROLIFERATION OF MATURE HUMAN CARDIOMYOCYTES DERIVED FROM INDUCED PLURIPOTENT STEM CELLS**	Julia María Halek (LIAN, INEU Fleni	No
	P4	Posters		
	Fecha y hora:	**21/11/2024 11:00-12:00 h**		
	Coordinadores:	**Alejandra Duarte, Mariana Fuertes, Marcelina Carretero**		
97	155	**LOW FREQUENCY GENES ASSOCIATED WITH HOLOPROSENCEPHALY**	Melisa Taboas (Centro Nacional de	No
99	385	**BIOCHEMICAL AND STRUCTURAL CHARACTERIZATION OF EXON13_15DUP VARIANT ASSOCIATED WITH FAMILIAL HYPERCHOLESTEROLEMIA**	Andrea Sánchez (Universidad de Cc	No
101	407	**FUNCTIONAL CHARACTERIZATION OF D47N-LDLR MUTATION ASSOCIATED WITH FAMILIAL HYPERCHOLESTEROLEMIA THROUGH EX VIVO ASSAYS**	Catalina Angélica Martínez Olea (Ur	No
103	438	**Analysis of variation spectra of 12 genes associated with thyroid dyshormonogenesis in a pilot cohort of children with Hyperthyrotropinemia**	Valentina Ricci (Centro de Investiga	No
105	493	**ROLE OF NR1I2 GENE VARIANTS IN ACUTE INTERMITTENT PORPHYRIA ONSET.**	Triviño Cuzzolino Facundo (Escuela	No
107	388	**APPLICATION OF AN OPPORTUNITY-BASED SCREENING FOR DETECTION OF PATIENTS WITH FAMILIAL HYPERCHOLESTEROLEMIA IN A HIGHLY COMPLEX HOSPITAL CENTER.**	Mariana Belén Opazo Torres (Facult	No
109	278	**GENETIC SCREENING AND BIOINFORMATICS ANALYSIS IN ARGENTINIAN PATIENTS WITH CONGENITAL HYPOTHYROIDISM**	Sebastián Rafael González (1Univer	No
	O1	Comunicaciones Orales		
	Fecha y hora:	**19/11/2024 16:00-17:00 h**		
	Lugar:	**Sala de Cámara**		
	Coordinadores:	**Liliana Rossetti, Ana María Buzaleh, Carina Rivolta**		
	154	**GENETIC CHARACTERIZATION OF 21-HYDROXYLASE DEFICIENCY COHORT BY LONG READ SEQUENCING**	Aldana Claps (Centro Nacional de G	No
	419	**DEEP PHENOTYPING AND NEXT-GENERATION SEQUENCING (NGS) IN CONGENITAL HYPOGONADOTROPIC HYPOGONADISM: DIAGNOSTIC YIELD IN ISOLATED AND SYNDROMIC CASES**	Lourdes Magdalena Correa Brito (Ce	No
	312	**RASOPATHIES: MOLECULAR DIAGNOSIS BY NEXT GENERATION SEQUENCING USING A CUSTOM GENE PANEL**	Paula Alejandra Scaglia (1 Unidad d	No
	357	**ANALYSIS OF ARRAY-CGH STUDY IN PATIENTS FROM ARGENTINA: PERIOD 2023-2024**	Julieta Laiseca (Centro Nacional de	No
	241	**Coexistence of two rare genetic diseases in patients with complex phenotypes revealed by Next Generation Sequencing**	María Esnaola Azcoiti (Unidad de M	No