Abstracts of poster presentations

Interdisciplinary poster session

Presentation number: IPS 1

CHALLENGES ASSOCIATED WITH MEDICAL TRAVEL FOR CANCER PATIENTS IN THE ARAB WORLD: A SYSTEMATIC REVIEW

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Each year, millions of patients around the world seek medical care abroad. Medical travel is becoming very common in the Gulf Cooperation Council Countries (GCC) due to many motivational factors. Cancer seems to be one of the top medical conditions for patients from the GCC to seek healthcare overseas. There are many factors associated with cancer patients seeking treatment overseas. However, unfortunately, there are very few studies that discuss the risks and challenges associated with the medical travel experience for those patients. We conducted a systematic review to summarize the evidence related to the complications and challenges associated with the medical travel experience for oncology patients in the Arab world. This systematic review was guided by PRISMA. PubMed was used as a search database by using a combination of medical travel, complications, and cancer keywords for publications which yielded 76 articles. Four coders independently determined eligibility based on PICOS and then extracted information from 14 articles. The resulting articles are based on three main categories, i.e., primary, and secondary data collection, and review articles. Of the total 76 articles, only 14 were included because they met the criteria. 62 articles were excluded because of irrelevance of the title, abstract, and insufficient data. Although this systematic review aimed to look at the medical complications that may arise from the medical travel experience for oncology patients, other challenges were found. The challenges reported can be grouped into the following themes: a) financial and economic aspects, b) medical care aspects, c) social and cultural aspects. Overall, more research studies are required in the Arab world for cancer patients treated overseas. The existence of such information around this topic will help in improving policies and strategies related to medical travel for the different stakeholders involved in the medical travel market. Moreover, these studies will not only aid in improving the quality of care for cancer patients who are engaging in medical travel, but they will also help in overcoming the challenges associated with medical travel experience for cancer patients at the different stages of the experience.

Key words: medical travel, treatment overseas, outsourced patients, treatment destinations, oncology

Presentation number: IPS 2

PRECISION MEDICINE AND CARDIOVASCULAR DISEASES: WHICH BIO JURIDICAL ISSUES MUST BE CONSIDERED?

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The area of cardio-vascular diseases - in translation from the animal to the human model - needs to test and validate many of the biomarkers identified, by demonstrating the presence/absence of a marker (be it a cDNA, a miRNA, a protein, a metabolite or a metabolomic profile) in healthy and affected tissue, that can be implemented in common clinical practice. Genetic variants were identified that modify the response to some relevant cardiovascular drugs, including beta-blockers (ADRB1, ADRB2, GRK5, GRK4); angiotensin converting enzyme inhibitors (ACE, AGTR1); diuretics (ADD1, NPPA, NEDD4L); and calcium channel blockers (CACNB2, CACNA1C). To date, however, genetic testing has not been employed routinely to guide selection of these drugs, and many genetic variants require confirmation in larger studies. The highlight topics can support the translational clinical application, with collaboration between knowledge that is the basis of precision medicine, in add taking evidence of unresolved bio juridical issues. The authors analyze some questions underlying precision medicine: equity in access preventive and therapeutic treatments based on pharmacogenetic principles; a guaranteed access to diagnoses and personalized treatments; the use of precision medicine diagnostics in the context of gender differences and its non-discriminatory use; use of precision medicine approach in prevention and its use in the workplace, with guarantees of the freedom and dignity of the person; integrity in the use of information located in biobanks. The guidelines proposed by scientific societies and National Institution of health are considered; the aspects worthy of discussion and bio-juridical implementation are emphasized. However fascinating, the area of interest of precision medicine, especially in the broad field of cardiovascular pathologies, also highlights some aspects that require careful evaluation and investigative criticism; therefore, requiring further regulatory interventions.

Key words: Precision Medicine, cardiovascular, genetic variants, biojuridical

Presentation number: IPS 3

AWARENESS REGARDING ORAL HEALTH AMONG ORTHODONTIC PATIENTS AND NON-ORTHODONTIC PATIENTS

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The aim of the study was to examine oral health awareness among orthodontically treated and orthodontically untreated patients, to examine their attitude about the importance of oral hygiene, their familiarity with toothbrushing techniques and familiarity and source of information on the use of toothbrushes and other toothpastes. Also, the aim was to examine their opinion on the importance of brushing teeth and their interest in expanding their own knowledge about oral hygiene. The study is organized as a cross-sectional study. Study included 98 patients of dental and orthodontic surgeries of the Health Center in Osijek. The examination was conducted during April 2020. An anonymous survey questionnaire was used to conduct the research. 98 patients participated in the study. Orthodontically treated patients use a toothbrush three times a day, far more than orthodontically untreated patients. They use a soft toothbrush and interproximal (interdental) toothbrushes significantly more than orthodontically untreated patients. They no longer use mouthwash, compared to orthodontically untreated patiens, but they go to regular check-ups much more than orthodontically untreated patients. Also, the orthodontist / dentist far more explains to them the importance of brushing teeth, which is not the case with orthodontically untreated patients. This study has shown that orthodontically treated patients have significantly more awareness regarding oral health and the importance of oral hygiene protocols, than orthodontically untreated patients.

Key words: oral hygiene, teeth, orthodontically patients, knowledge, attitudes

Presentation number: IPS 4

CONTRADICTORY ARGUMENT OF HYPERTENSION AMONG YOUTHFUL AGES

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Hypertension is one of the major community health challenges among elderly people on the risk of cardiovascular disease, is now a disease that is found among people who fall within the age brackets between 20 and 30 years. Our community outreaches reveal this at Ikorodu and Lagos Mainland Local Governments in particular. A community cross-sectional research was carried out and it was shocking that people of the aforementioned age brackets were found hypertensive during one of our Community Medical Outreaches at the aforementioned Local Government Areas. Our discovery was terrific that the prevalence of hypertension was found among males than females during the BP test to found results as 125.27 ± 17.08 mmHg and 93.55 ± 9.48 mmHg, respectively. TV-TAY Wellbeing Initiative has a pre-conclusive agreement to present this that the odds of being hypertensive in the community is majorly the problem of challenges such as economic instability the situation, social unrest, and political disenchantment in the country which bring about the unnecessary taking of uncontrollable alcoholic drink, tobacco, illiteracy unawareness of the danger with continuous indulging and engagement because of unawareness risk of it based on our findings.

Key words: uncontrollable alcoholic drink, tobacco, illiteracy unawareness, hypertensive

Presentation number: IPS 6

ASSOCIATION BETWEEN POLYGENIC RISK SCORES FOR PLASMA PROTEIN N-GLYCOSYLATION TRAITS AND 273 ICD-10 DISEASES

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N-glycosylation is a post-translational modification of proteins by covalent attachment of a carbohydrate structure to an asparagine residue. N-glycans physical properties and biological activity of the serum proteins. The composition of the N-glycome of blood plasma proteins is changing in ageing and diseases, therefore, N-glycans are often regarded as promising biomarkers of various physiological and pathologic states. Recently, genome-wide association studies (GWAS) identified a number of loci that are involved in regulation of plasma protein N-glycosylation. Some of these loci are also associated with inflammatory, autoimmune, cancer and other diseases. Elucidating the degree of genetic architecture overlap and causal relationships between plasma N-glycome and diseases, remains one of the important tasks of the glycobiology field. To answer this question, we have used polygenic risk score (PRS) analysis. We have calculated PRSs for 117 plasma N-glycosylation traits using summary statistics of a GWAS performed in a cohort of European descent (N = 7541), where 31 loci were found to be associated with N-glycosylation. As a next step we are planning to perform an association study of the PRS for the 117 plasma glycan traits and 273 diseases included in the 10th revision of the International Statistical Classification of Diseases and Related Health Problems (ICD-10) list, available through the UK BioBank (UKBB) database, with prevalence > 0.05 and < 0.95. For the pairs of glycan trait and disease that show a statistically significant association we plan to perform Two-sample Mendelian randomization (MR) analysis in both directions to answer the question of causal relationships between the said traits. As exposure and outcome for the Two-sample MR we are going to use summary statistics from GWAS for plasma N-glycosylation traits performed in a cohort of European descent (N = 7541) and those available from UKBB for the diseases. We expect our research project to shed light on the connection between plasma protein N-glycome and various pathologies, on the genetic basis of protein N-glycosylation, to improve the existing glycan biomarkers and potentially to discover new ones. Acknowledgments: This work was supported by a grant from the Russian Science Foundation (RSF) No. 19-15-00115.

Key words: N-glycosylation, plasma proteins, polygenic risk scores, Mendelian randomisation

Presentation number: IPS 7

SEX DIFFERENCES IN RISK OF BLOODSTREAM AND SURGICAL SITE INFECTIONS

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Due to the exponential increase in the incidence of nosocomial infections and the increasing rates of antibiotic resistance, the identification of nosocomial infections' risk factors remains a current topic. Sex is one of the risk factors currently under study. The role of sex in the context of urinary tract infections (UTI) is well known. Conversely, the link between sex and bloodstream (BSI) and surgical site (SSI) infections is less well known and more uncertain. the author analyzed the literature data about the role of sex in the BSI and SSI development. significantly higher BSI and SSI incidence rates were observed in the male population. It has been hypothesized that it may be due both to genetic factors related to the thicker and coarser hair in men, and to the shaving of the hair. This hypothesis is further supported by the absence of sex differences with regard to the incidence of BSI and SSI in the pediatric population. Conversely, in the adolescent population the girls had significantly lower odds of community-associated BSI and lower odds of SSI. Biological differences between men's and women's skin could constitute another possible reason for gender differences in the case of BSI and SSI incidence. A greater bacterial colonization of the skin surrounding the surgical insertion site of the central venous catheter was observed in men compared to women. Furthermore, sex would affect the immune system's response via the hormonal system. In fact, the levels of estrogen promote the response of the immune system, playing a protective role; differently androgens suppress it. The increased rates of infection in the female population observed during menopause would confirm these observations. It has also been hypothesized that women may be at greater risk for SSI in the case of cardiac surgery due to the small size of the arteries and the increased tension of the thoracic incisions due to the pendulous breasts. New studies are expected regarding cultures of the bacterial colonization of the skin surrounding a central venous catheter at the insertion site or a surgical wound to clarify the role of genetics and sex in the development and incidence of BSI and SSI in order to customize the preventive and therapeutic strategies of BSI and SSI and reduce their mortality.

Key words: sex, risk infection, surgical site infection, bloodstream infection

Abstracts of the interdisciplinary session of American academy of forensic sciences and ISABS 2022

Presentation number: AAFS 1

FENTANYL: HISTORY, ABUSE, DANGER

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Illegal drug use and trafficking in the United States is at an all-time high. In addition to traditional drugs of abuse (Heroin, Cocaine, Methamphetamine and Marijuana) there has been a huge increase in the abuse of Novel Psychoactive Substances (NPS) such as Fentanyl during the last 15 years. Illicit abuse of Fentanyl and Fentanyl-related synthetic opioids began in the mid 1990's of in the United States. This was the result regulations, policies and medical practice that focused on opioid medications (i.e. Oxycodone) as the primary treatment for pain. Hospital management and administration encouraged doctors to reduce patient pain to a self-described level of zero. Pharmaceutical manufacturers were eager to provide the products. As patients became addicted they had difficulty getting prescriptions legally renewed and then sought other sources for similar drugs. Gradually, other supply networks developed to provide Oxycodone and look-alike clandestinely manufactured synthetic opioids. Eventually, US drug control regulations improved, which led to entirely new substances of abuse. Fentanyl became a popular drug of choice. Precursor chemicals originate in China that are then shipped to Mexico for synthesizing into Fentanyl and Fentanyl analogues. These products are commonly mixed with Heroin, Cocaine, Methamphetamine, Marijuana and also compressed into look-alike tablets such as XANAX. It should be noted that the illicit drug user and the drug trafficker frequently don't know the content or the concentration/potency of the drugs. There are many literature references that refer to the abuse of Fentanyl as like playing "Russian Roulette'. Fentanyl is 50 times more potent than Morphine and 100 times more potent than Heroin. Fentanyl analogues are even more potent. In 2021 there were 100,000 Fentanyl related drug overdose DEATHS in the United States. What about the future?? It might get worse before it gets better. Has anyone heard about Isotonitazine compounds?

Key words: Fentanyl, Synthetic Opioids, Drug Abuse

Presentation number: AAFS 2

COLLABORATION TO EFFECT IDENTIFICATION FOR UNKNOWN INDIVIDUALS RECOVERED IN A FORENSIC CONTEXT

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In 2015 Arizona State University and the Maricopa County Office of the Medical Examiner agreed to collaborate to advance identification efforts for badly degraded and burned decedents. The concept was to apply techniques derived for extracting and amplifying ancient DNA samples to these individuals to assess the efficacy for obtaining useful profiles. In Arizona, as in many arid, hot environments, individuals who die in the desert and are not found for a protracted time are subjected to extremes of temperature, solar exposure and animal predation. These combine to remove the organic matrix of the bone and the most likely sources of usable DNA. Current analytical kits have failed to produce enough DNA for scientific identification. Similarly, in badly burned bodies there is a threshold beyond which standard techniques cannot extract enough DNA to create a strong profile. One mandate of a medical examiner or coroner is to make an identification of decedents within their jurisdiction. When standard techniques are unsuccessful, the decedent remains unidentified. Practitioners are constantly reviewing these cases in the context of scientific advances and thinking outside of the box to effect opportunities for identification. The proposal to use aDNA methodologies for these cases provides a new avenue for success. NIJ funding has allowed testing several methods in order to determine which is the best for yielding enough DNA for identification. The protocols have had differing levels of success ranging from very good to no results. Predictably the cremated and most degraded samples are the ones with minimal yields. DNA profiling is a boon to forensic science in many contexts. Identification of unknown decedents is a small portion, but it plays a critical role. Without identification families are left in limbo, legal processes stop, and law enforcement investigations are derailed. In the United States there are tens of thousands of unidentified decedents languishing in medical examiner and coroner offices, or in pauper cemeteries. Advancing methods for assessing these individuals creates hope that more will move from the unidentified column into the identified column. This research hopes to play a large role in that transition.

Presentation number: AAFS 3

THE PAST, CURRENT, AND FUTURE OF FORENSICS IN THE US. CRIME SCENE TRAINING

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Contemporary law enforcement has expanded its ability to solve crimes by the adoption of standardized forensic testing procedures. Today, crimes can only be solved by the combinations detailed examination of the crime scene and analysis of forensic evidence, the utilization of the open and close sources of big data bases and the theory of Cloud data mining and crime scene reconstruction. Knowledge of forensic evidence is not only crucial in criminal investigations and prosecutions, but also vital in civil litigations, major man-made and natural disasters, and the investigation of global crimes. However, there was not much consensus on what and how to be training or educate future forensic scientists in crime scene investigation. In the past, this type training is done by localized apprenticeship. The current training program is focused on the standardized procedure for crime scene technician. This type of inadequate training was unfortunately overlooked and created many issues and problems for forensic field. The future crime scene training program should be conducted with a global effort in the ability of observation, and recognition, the systematic and logical analysis, the correct interpretation of test results, and reconstruction of the scene. This includes not only the first responding officer, detective, the crime scene technicians, investigators, forensic scientists but also the judges and attorneys. The training of advanced investigative skills (such as GPS positioning, Cell phone tracking, Social network, E-evidence, Video image analysis, Big Date Analytics, Artificial intelligence, Theory formation, Forensic DNA Genealogy, Trace Evidence Analysis, Pattern Evidence Recognition and Crime Reconstruction) will be discussed, and the ability to observe and analysis of crime scene and Forensic Evidence will also be covered, Famous cases will be utilized to illustrate the importance of new concepts in forensic crime scene training.

Presentation number: AAFS 4

FORENSIC SCIENCE STANDARDIZATION EFFORTS BY THE AMERICAN ACADEMY OF FORENSIC SCIENCES (AAFS) ACADEMY STANDARDS BOARD (ASB)

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The goal of this presentation is to educate an international audience on the history and progress and also the motivation behind the American Academy of Forensic Sciences, AAFS, Academy Standards Board, ASB. The presenter is the past president of the Academy with extensive experience with standards development organizations. The AAFS' contribution to forensic science standards has been significant since first forming its standards development organization beginning in 2015. In that same year it received American National Standards Institute, ANSI, accreditation for the American Standards Board, the ASB. Material and Methods – Academy Standards Board, ASB, statistics on standards, guidelines, and technical reports were utilized to present a historical account of ASB progress. When the ASB was created, consensus bodies were formed representing numerous forensic science disciplines. Within the consensus bodies, the interest categories, developed and approved by ANSI, represent various stakeholders who provide various perspectives resulting in standards that meet necessary objectives by the end user. A public balloting process, an ANSI requirement, ensures a free and open process. Results – The presentation will include types of interest categories, stakeholders, ASB procedural requirements, and up to date progress and goals of the ASB consensus bodies. Since the date of inception, the ASB has gone from an idea to a fully functioning development program with volunteers from thirteen (13) forensic disciplines creating over 76 standards, guidelines, or technical records. Conclusion – The ASB 's progress in publishing standards, guidelines, and technical reports is impressive given the relatively short period of time since they were formally established and accredited by ANSI. That progress, along with the history of the program, will be provided in detail in this presentation.

Key words: AAFS, ASB, forensic standards

Presentation number: AAFS 5

CORRECTING MISCARRIAGES OF JUSTICE – INNOCENCE PROJECT OF CROATIA

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The presentation focuses on the issue of wrongful convictions and subsequent DNA testing as a method for proving innocence at persons deprived of liberty, through the perspective of the newly established and experimental “Innocence Project in Croatia”. The presenters will talk about the project’s main aim, specific objectives, as well as obtained initial results from the field research. Furthermore, the presenters will present the positive influence of the American innocence projects of the Croatian criminal justice system, through the perspective of the Croatian legal practice and the position of the criminal justice system as a whole concerning the ramification, obtaining and storage of DNA evidence from severe form of criminal acts, including the re-opening of the criminal procedure due to new DNA evidence as a cause wrongful conviction. The presentation will conclude with the encouragement of establishing a national database providing detailed information on previous cases of wrongful convictions and the positive experiences from the U.S. National Registry of Exonerations and the establishment of conviction integrity units as positive examples from which Croatia and other legal jurisdictions in the region should learn.

Presentation number: AAFS 6

ARREST-RELATED DEATH BY PRONE RESTRAINT CARDIAC ARREST

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Several causes of death have been postulated for agitated subjects who have succumbed after a police encounter, particularly when they have been restrained in a prone position. Much of the literature has focused on excited delirium or restraint asphyxia as the cause of these deaths; often stress with underlying cardiac issues has been imputed. A recent review of the literature declares that restraint is the common factor.[1] More recently, Steinberg ascribed acute severe metabolic acidosis with an inability to compensate with respiratory alkalosis as the cause of these deaths in the setting of prone restraint and proffered the term, “Prone Restraint Cardiac Arrest” (PRCA).[2] Pulseless electrical activity (PEA) and asystole are consistent with this concept. We present two cases that demonstrate the danger of prone restraint and, we believe, serve as examples of PRCA. The first case involved an obese 51-year-old male subject, high on PCP and cocaine and hallucinating. His hands were cuffed behind his back and his legs shackled by police while supine on the ground. Emergency medical personnel placed him prone and strapped on a manual stretcher, which was then placed on a wheeled stretcher and further straps placed. He struggled against the restraints, but then became unconscious as he was loaded on the ambulance. He was repositioned supine in the ambulance. He had been prone for 10 minutes. His initial EKG rhythm was bradycardia, but subsequently no carotid pulse was felt and then the electrical activity was lost (asystole). He arrived in the emergency room in a state of pulseless electrical activity (PEA), but he was successfully resuscitated. His end title CO_2 was found to be markedly elevated. Upon his hospital admission, an arterial blood gas revealed severe acidosis (pH of 7.015) and a significantly elevated pCO_2 (70.5 mmHg). An echocardiogram in the hospital showed a normal heart function. He died of brain death 3 ½ days after his arrest. Autopsy revealed an enlarged and dilated heart with no gross atherosclerosis and evidence of reperfusion injury. The second case involved an obese 41-year-old male, high on amphetamine and methamphetamine, seen by police stepping in and out of a busy roadway, shouting and talking to an imaginary, threatening person. He sat, sweating and breathing heavily, as requested by the police and his hands were cuffed behind his back. He suddenly got up and began to run when three officers took him to the ground where he thrashed about and kicked his legs. Police rolled him prone and leg shackles were applied. At one point, he uttered, “I can’t breathe!” Emergency medical personnel arrived and placed a rigid board on his back and an officer was asked to sit on top. After a couple of minutes, he was turned supine and found to be unresponsive. He had been prone for 15 minutes. ACLS was initiated. The EKG revealed PEA which deteriorated to asystole. Venous blood gases revealed a profound acidosis (pH 6.64) and a markedly elevated pCO_2 (157 mmHg). He was pronounced dead shortly after arrival in the emergency room. An autopsy disclosed bruising on his back, a moderately enlarged heart with focal severe coronary artery stenosis.

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Presentation number: AAFS 7

OPEN WATER AQUATIC SCENE PROCESSING FOR DEATH INVESTIGATIONS

Andrea Zaferes

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Learning Overview: After attending this presentation, attendees will understand the critical need of crime scene investigators (CSI) and other investigators to identify, document, and preserve potential evidence on open water scenes. Impact on the Forensic Science Community: This presentation will impact the forensic science community by demonstrating the unique challenges presented by bodies reportedly found in open water scenes, the detection and documentation of evidence in open water scenes, and the recognition and investigation of crimes that involve open water scenes. This presentation will present novel solutions, tools, and protocols to utilize CSIs, marine patrol officers, public safety dive team personnel, and other first responders, to identify and collect often-missed evidence. Bodies reportedly found in open water (BRFIOW) may represent natural, accident, suicide, and homicide manners of death. In the latter case the death may have occurred on land with a postmortem body disposal, may have initiated on land with resulting incapacitation followed by submersion with one of the causes of death being drowning, or with the homicide solely involving submersion and drowning. Asphyxiation, whether criminal, accidental, or suicidal, is a process that can be fatal or non-fatal. Asphyxiation directly impedes respiration or blood flow to the brain through various methods, such as strangulation (1), drowning (2), and inert gas (3). Criminal asphyxiation occurs in domestic violence (4), child/elder abuse (5), sex crimes (6), state and non-state tortur (5), and human trafficking (7). The diagnosis of fatal asphyxia, criminal or benign, requires an informed and accurate scene investigation, and a careful exclusion of underlying injury or disease processes (8). Despite the adoption of widespread legislation(x) specifically targeting asphyxiation violence, evidence collection is sparse and convictions are rare (9). This is partly because criminal asphyxiation often presents with nonspecific findings (10) and minimal or no obvious external injury, can be a diagnosis of exclusion, and lends itself to crime scene staging (11). Therefore, these cases require the preservation and collection of often circumstantial evidence present at the initial scene. Capturing this evidence is especially critical when victims and witnesses may be unable to participate beyond the initial scene for fear of retaliation and safety. As such, crime scene investigators (CSIs) are in a unique position to provide a variety of solutions. It is the job of CSIs to document and process a scene (location, victim, suspect) in its totality as it appears upon arrival. In criminal asphyxiation cases, CSIs are the most apt to capture often missed evidence, including fingernail scrapings and injuries present on suspects. CSIs may also be the only practitioners who can forensically document initial and follow-up injury presentation. For example, in non-sexual asphyxiation cases, victims are unlikely to receive forensic nurse examinations. CSIs' interactions with victims and suspects can also provide unique opportunities to identify and document signs and symptoms of asphyxia and indicators of abuse. For equivocal fatal cases, CSIs can provide the information necessary for investigators to detect staged scenes and for medical examiners to accurately determine cause and manner of death. This novel use of CSIs can be achieved by developing protocols, using investigative forms, and implementing training. Protocols are needed to dispatch CSIs to possible criminal asphyxiation scenes and to provide guidance for evidence recognition, documentation, and processing. CSIs must also be properly equipped with knowledge and evidence-based forms in order to effectively work these often-challenging cases. A current obstacle, as will be demonstrated in this presentation, is insufficient CSI training on criminal asphyxiation in the US as shown by a lack of its mention

in CSI textbooks and training syllabi. Overcoming this requires a forensic community response. As a first step in this response, this presentation will introduce tools for CSIs, including investigative forms and protocols for possible asphyxiation scenes. This presentation illustrates the challenges presented by criminal asphyxiation, provides solutions that CSIs can provide, and CSI tools for investigating strangulation cases. With standardized agency protocols, tools, and specialized criminal asphyxiation training, CSIs can help identify, document, and preserve critical evidence in these cases.

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Abstracts of 8th Croatian human genetics conference & 1st Croatian personalized and precision medicine conference

Presentation number: CSHG 1

ACETABULAR PROTRUSION - UNDERESTIMATED BUT FREQUENT DEFORMITY IN PATIENTS WITH OSTEOGENESIS IMPERFECTA

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Acetabular protrusion (AP) is common deformity in people with osteogenesis imperfecta (OI) or brittle bone disease, a genetic disease of the connective tissues caused mainly by mutations in collagen type I. Clinical features of OI are brittle bones, low-energy fractures, skeletal deformities, joint laxity, blue sclerae, dentinogenesis imperfecta, cardiovascular and respiratory problems and hearing loss. Research on AP in OI is limited. However, AP is found in about 50% of people with OI, with even increase to 70% in people with OI type III. The purpose of this text is to draw attention to this frequently unrecognized deformity. We reviewed recently published articles in orthopaedic literature and compared data from literature with data on patients with OI treated in our hospital in the last five years (2016. – 2021.). Radiographs of hips and pelvis in our 17 patients (10 females and 7 males) were reviewed. Literature review has shown that AP is predominantly responsible for gastrointestinal problems and for increased risk for proximal femoral fractures and particularly for femoral neck fractures in OI population. We found one patient with OI type I, thirteen patients with OI type III; two patients with OI type IV and one patient with X-linked type OI. In our cohort, 65% of our patients with OI type III had AP of some degree. Three adult patients had significant AP as measured according to Kohler line. Two of them had obstruction constipation as gastro-intestinal complication of AP and this was resolved with dietary adjustments. No patients had femoral neck fracture, so far. This could be explained that majority of our patients were children and had limited ambulation prior long bone corrective surgery with expandable intra-medullary implants. There are single case reports of technical surgical problems in urologic prostate surgery due to AP in adult patient with OI type I, as well as carcinoma of left colon presenting as mechanical obstruction in a patient with osteogenesis imperfecta type III. In multidisciplinary management approach for patients with OI one should not underestimated deformity of pelvic bones, AP in particular, as there are significant potential problems that can affect quality of life in this population.

Key words: osteogenesis imperfecta, acetabular protrusion, treatment, prevention

Presentation number: CSHG 2

THE ROLE OF PHARMACOGENOMICS IN EVALUATING THE EFFICACY AND SAFETY OF DRUGS

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To achieve a pharmacological activity, drugs should undergo passage through cell membranes from the site of application, transport to cell compartments, biotransformation to active or inactive metabolites, elimination from the body, and finally bind to specific biological macromolecules (molecular targets). The biological molecules involved are metabolic enzymes and transporters, membrane/cell receptors, and other intra- or extracellular proteins. The polymorphisms of the genes encoding these proteins can lead to changes in the drug's pharmacological effect, resulting in therapeutic failure and/or the occurrence of adverse drug reactions. Variations in genes involved in drug absorption, distribution, metabolism, and elimination (ADME) can alter its pharmacokinetic profile, influencing systemic exposure and concentration at the site of action. Additionally, variations associated with a drug's target molecules can directly affect its pharmacodynamic effect. In the last twenty years, pharmacogenomics has attracted attention as a discipline that can contribute to the quality of patient health care and present an essential part of the personalized medicine concept. The use of genetic information would help predict the response to the drugs to enable safer, more effective, and cost-effective treatment to each patient.

Presentation number: CSHG 3

INFLUENCE OF GENETIC SUB-STRUCTURING OF STATISTICAL FORENSIC PARAMETERS ON GENETIC STR MARKERS IN THE POPULATIONS OF SOUTHEASTERN EUROPE

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The goal was to perform a meta-analysis of synthesized data and investigate the influence of specific intrapopulation genetic structures on interpopulation relationships. Special focus was the influence of island population isolation on the substructuring of the Croatian population, and the influence of regional population groups on the substructuring of Southeast Europe. A long-term goal is to develop a model of appropriate sampling of the total population when creating a database of genetic STR markers that would properly reflect all the characteristics of included subpopulations. Autosomal STR loci were analyzed using four forensic parameters (match probability, power of discrimination, power of exclusion and the degree of polymorphism) on a sample of 2877 unrelated participants of both sexes. The analysis was performed using the statistical package PowerStats v1.2. The comparison of forensic parameters between different subpopulations of Croatia and Southeast Europe indicates that the isolation of individual Croatian subpopulations and certain rare alleles in their gene pool affect the values of forensic parameters. Specific features of (sub)populations should be taken into account for appropriate sampling of the total population when creating a DNA database of STR markers.

Key words: STRs, genetic sub-structuring, forensic parameters, Croatia, Southeast Europe

Presentation number: CSHG 4

EXOME SEQUENCING AND AUTISM SPECTRUM DISORDER- DIAGNOSTIC CHALLENGES AND FUTURE DIRECTIONS

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The genetic basis of autism spectrum disorder (ASD) is still poorly understood in many patients. Next-generation sequencing enables simultaneous detection of pathogenic variants in hundreds of disease-causing genes. The goal of this study is to define diagnostic utility of clinical exome sequencing in elucidating autism spectrum disorder (ASD) etiology and create future diagnostic and therapeutic directions. Material and Methods For this study, we analyzed 55 ASD patients that were diagnosed and treated at the Department of Medical Genetics and Reproductive Health in Children's Hospital Zagreb. All of them had a confirmed ASD diagnosis and were underwent to detailed clinical geneticist's evaluation. Chromosomal disorders and fragile X syndrome have been previously excluded in all patients. Clinical exome sequencing has been performed using Illumina TruSight One Kit. In 14 out of 55 patients (25.4%) pathogenic variants were identified (14/55) and involved genes: CAMTA1, DEAF1, EP300, DICER1, MED13, CHD7, DYRK1A, FOXG1, SOS1, MED12, EHMT1, CHD8, TCF4, NFIX, PAK3. Variants of unknown significance (VUS) were present in 16.4 % of patients (9/55), in genes: AUTS2, DLG3, IHIF1, DEAF1, BICRA, CREBBP, CTNNB1, RELN. The remaining patients had negative test results (32/55; 58.2%). Clinical exome sequencing elucidated genetic basis of autism in 25 % of ASD patients, mostly attributable to genes involved in fundamental genetic processes; transcription (CAMTA1, DEAF1, EP300, MED13, FOXG1, MED12, TCF4, NFIX), chromatin remodeling (CHD7, CHD8, EHMT1), ribonuclease activity (DICER1) and activity of various kinases (DYRK1A, SOS1, PAK3). High percentage of negative and inconclusive (VUS) results (74.6%) requires additional genetic (whole exome/genome), epigenetic and environmental risk factor analysis. Establishing autism genetic basis is the prerequisite for the development of new therapeutic strategies and personalized treatment for the ASD patients. Acknowledgment: This work was supported by Scientific Center of Excellence for Reproductive and Regenerative Medicine and by the EU through the European Regional Development Fund, under grant agreement No. KK.01.1.1.01.0008, project „Reproductive and Regenerative Medicine - Exploring New Platforms and Potentials"

Key words: autism, exome sequencing, personalized management

Presentation number: CSHG 5

SEX-RELATED IMMUNOPHENOTYPE DIFFERENCES IN HUMAN STROMAL VASCULAR FRACTION FROM LIPOASPIRATE AND MICROFRAGMENTED LIPOASPIRATE REVEALED BY POLYCHROMATIC FLOW CYTOMETRY

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Mesenchymal stem/stromal cells or from recently referred to as medicinal signaling cells (MSC) hold tremendous therapeutic potential in regenerative medicine. Although successfully used for treating knee osteoarthritis (OA), a broader application of MSC in the field requires a better understanding of functional and cellular heterogeneity. In order to gain insight into the human MSC from adipose tissue applied for autologous OA treatment, we performed extensive comparative immunophenotyping of stromal vascular fraction (SVF) from lipoaspirate or microfragmented lipoaspirates by polychromatic flow cytometry. Sixteen OA patients (eight females and eight males) were enrolled in the study. For each of the patient SVF was obtained from lipoaspirate and microfragmented lipoaspirate counterpart. Isolated SVF cells were stained with fluorochrome-labelled antibodies specific for the CD31, CD34, CD45, CD73, CD90, CD105, CD146 cell surface markers and analyzed by flow cytometry. We found an enrichment of the endothelial progenitor cells in the clinically applied microfragmented stromal vascular fraction. Sex-related differences were observed in the MSC marker expression and the ratio of the progenitor cells from fresh lipoaspirate; in female patients it contained a higher expression of CD90 on the three progenitor cell types including pericytes, a higher expression of CD105 and CD146 on CD31highCD34high endothelial progenitors as well as of CD73 on supraadventitial-adipose stromal cells. Some of these MSC-expression differences were present after microfragmentation,

which indicates a diverse phenotype pattern of the applied MSC in female and male patients. Adding to the understanding of the perplexed heterogeneity of the adipose MSC subpopulations serving as OA therapeutics, our results tackle on the sex-related molecular signatures in women and men that might contribute to a personalized approach of regenerative medicine.

Key words: osteoarthritis, mesenchymal stem/stromal cells, medicinal signaling cells, stromal vascular fraction, immunophenotyping

Presentation number: CSHG 6

CROATIAN GENETIC HERITAGE: RENEWED Y CHROMOSOME STORY TWO DECADES LATER

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The aim of the study was to analyze additional set of Y-Chromosome genetic markers to acquire a more detailed insight into the diversity of the Croatian population. The total number of 518 Yfiler™ Plus profiles was genotyped. Allele, haplotype frequencies and haplotype diversity, were calculated using the STRAF software package v2.0.4. Genetic distances were quantified by Rst using AMOVA online tool from the YHRD. The evolutionary history was inferred using the neighbor-joining method of phylogenetic tree construction in MEGAX software. Whit Athey's Haplogroup Predictor v5 was used for additional comparison with selected European populations. The total of 507 haplotypes were used for genetic STR analysis. The interpopulation comparison with the original 27 Y-STR markers shows the lowest genetic diversity between Croatian and Serbian population, and the highest between Croatian and Spanish population. Interpopulation study on 17 Y-STR markers shows the lowest genetic diversity between Croatian and Bosnian-Herzegovinian population, and the highest between Croatian and Irish population. Total of 518 haplotypes were used in the determination of haplogroup diversity. Haplogroup I with its sublineage I2a expressed the highest prevalence. Haplogroup R, with its major sublineage R1a, is the second most abundant

in the studied Croatian population, except for the subpopulation of Hvar, where E1b1b is the second most abundant haplogroup. Rare haplogroups also confirmed in this study are L, T and Q. G1 is detected for the very first time in Croatian population. New insight into differences between examined subpopulations of Croatia and their possible (dis)similarities with neighboring abroad populations was notified.

Key words: Y chromosome, haplogroup, Croatian population, STR, genetic heritage

Presentation number: CSHG 7

CHROMOSOMAL MICROARRAY IN CLINICAL DIAGNOSIS OF CEREBRAL PALSY

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Cerebral palsy (CP) is a group of non-progressive disorders of posture, tone, and/or movement. It is caused by a non-progressive lesion of the developing brain or brain malformation. Recent studies have implicated that genetic factors could contribute to diagnosis of CP or even cause CP. Potentially deleterious genomic copy number variants (CNVs) have been found in several CP cohorts, but estimates varied considerably depending on study criteria. The goal of this study is to define diagnostic utility of chromosomal microarray in children with CP, with well-defined phenotype according to Surveillance of cerebral palsy in Europe (SCPE) criteria. This study included 79 patients with CP, referred to the Department of Medical Genetics and Reproductive Health, Children's Hospital Zagreb. All of them had confirmed CP diagnosis by the criteria of SCPE. The analysis was conducted using Agilent 60K oligonucleotide array-based comparative genomic hybridization. Clinically relevant variants were detected in 7 of 79 patients (8,86%): deletions 14q32.31q32.33, 1q21.1q21.2, 15q11.2, 17p13.3 and 22q11.2; duplications 1q21.1q21.2 and Xq28 (in male patient). Variants of unknown significance (VOUS) were present in 5 patients (6,33%): duplications 15q11.2, 3p26.3p26.2, 18p11.31 and Xq28 (in female patient) and deletion 17p12. Remaining patients had negative test results. Among the patients with clinically important variants, three had brain MRI classified as maldevelopment, three as predominant white matter injury, and one patient with 22q11.2 deletion as predominant grey matter injury. The genomic architecture of CP is highly complex, similar to other neurodevelopmental disorders. Continued analysis and reporting of CNV findings alongside massively parallel SNV analyses are needed to expand our knowledge of CP. Better understanding of all the possible genes involved in CP etiology is the cornerstone for understanding neurobiology of CP. This work was supported by Scientific Center of Excellence for Reproductive and Regenerative Medicine and by the EU through the European Regional Development Fund, under grant agreement No. KK.01.1.1.01.0008, project „Reproductive and Regenerative Medicine - Exploring New Platforms and Potentials".

Key words: cerebral palsy, chromosomal microarray, copy number variants

Abstracts of the anthropological session “Bioanthropology and global health in the times of crisis”

Presentation number: ANTR 1

INTEGRATIVE APPROACHES TO GLOBAL HEALTH

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Modern bioanthropology, although always interdisciplinary, nowadays is breaking down barriers in science ever more efficiently. Multiple factors – genetics, environment and lifestyle – influence disease development. The main problem of multiple branches of science is their usual monochromatic perspective, where every problem is viewed from its exclusive point of reference. Our genetics is a result of natural selection, and although we are as fit as can be, we are the result of all the adaptations that human species acquired during the past, not only as our species, but also as all our ancestral species. Furthermore, with the advancement of science and technology (reflecting in every aspect of our everyday life), human lifestyle has dramatically changed in a brief amount of time on a historical timescale. Unfortunately, adaptive changes of the genome are not able to compensate and, as a result, multiple non-communicable diseases (e. g. diabetes, cardiovascular diseases, tumours) are emerging within human populations worldwide with increasing costs for the health systems on a global scale. In order to decrease the incidence of diseases and consequently their financial and social burden, a comprehensive approach taking into account social, behavioural, psychological, and biological factors is necessary. By applying methods from analytical chemistry, molecular biology and biochemistry combined with different types of questioners and interviews and data from populations from the past, we can better understand causes of diseases, both on organism and molecular level. Importantly, taking into account environment (toxicological burden) and ecological systems inhabiting our organisms (microbiota) bioanthropology can strongly contribute to finding the proper treatment of diseases in personalized manner and, more importantly, help in their prevention.

Presentation number: ANTR 2

TOXICOLOGY IN THE CONTEXT OF GLOBAL HEALTH

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Human health has been immensely influenced by environmental factors. Various reports have shown that nearly 25% of global deaths and about a third of the world’s burden of disease in children can be attributed to such factors. Although some environmental contaminants are difficult to avoid, amelioration or even elimination of these factors is possible and the diseases arising because of these factors are preventable. Accordingly, this attributable disease burden is often 10-fold higher in poor countries than in developed ones. This disparity is mainly caused by a lack of modern technology, weak laws and regulation in the field, a lack of awareness among the population, and general poverty in these countries. Furthermore, residents of wealthy countries are also affected by many environmental factors such as air pollution, lead poisoning, etc. Hence, the role of environmental health in global health is of great concern. The most potent environmental causes of worldwide illness are unclean water, air pollution, and exposure to various industrial chemicals. Concerning chemical exposure, it is important to stress that of the more than 30,000 chemicals commonly used today, scientists have studied only fewer than 1% in detail, while our understanding of the effects of simultaneous exposure to low levels of hundreds or thousands of chemicals we face on daily basis is rudimentary, at best. Cohesive and comprehensive policies that would protect both people's health and planet health require strengthening of the institutional capacity and social actions that encompass raising public awareness and public health measures in the field.

Presentation number: ANTR 3

CHILD GROWTH AND ARMED CONFLICT

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The widely publicised invasion of the Ukraine by Russia has highlighted yet again the devastation caused by armed conflict at individual, community, and national levels. Human society in the 20th and 21st Centuries has never been free of armed conflict either within or between nations. Whilst the first half of the 20th century was dominated by the 'World Wars' of 1914-18 and 1939-45, the Uppsala Conflict Data Program records that never less than 40 countries worldwide were involved in armed conflicts between 1989 and 2018. In their Lancet review of 2021, Bendavid et al (2021) emphasise that whilst "…every conflict-affected region, every conflict, and every affected community is different from all others…" common features are shared and yet the evidence that supports health consequences is "weak" and "limited", and in the case of adolescents almost "non-existent". The status of human growth and development over time has been used globally to determine the health and wellbeing of children. Given the ubiquitous nature of armed conflict, it is not surprising that research in human growth and development has provided several significant studies highlighting the effects of armed conflict on children and adolescents in war zones. Data reflecting the effect of the 1st and 2nd World wars on young people in England, the Netherlands, Germany, and Japan, demonstrates the power of armed conflict to interrupt the positive secular trends characteristics of improved physical growth and development at national levels in both those experiencing conflict first hand, and the long-term consequences to those experiencing it through maternal exposure during pregnancy. Studies of the growth outcomes of armed conflict are, of course, complicated by the inability to plan prospective studies. Most studies examine proximal outcomes of conflict such as mortality, malnutrition, injury, disability, and disease. However, the existence of repeated cross-sectional surveys provides unequivocal retrospective evidence for the reduction of growth rates across the age range from infancy to adulthood and the differential effects of armed conflict in children and adolescents.

Presentation number: ANTR 4

UNRAVELLING DATA FOR RAPID EVIDENCE-BASED RESPONSE TO COVID-19 – INANTRO EXPERIENCE FROM THE UNCOVER PROJECT

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"unCoVer-Unravelling data for rapid evidence-based response to COVID-19" is a Horizon 2020-funded network of 29 partners from 18 countries. Formed with the aim to collect and use real-world data (RWD) resulting from response and provision of care to COVID-19 patients by health systems across Europe and elsewhere, unCoVer exploits the full potential of this information to rapidly address clinical and epidemiological research questions arising from the COVID-19 pandemic. These heterogeneous datasets comprise from information on over 22 000 hospitalized patients, as well as registry data on over 1 900 000 COVID-19 cases across Europe, with continuous updates. So far, the datasets have been described, harmonized and integrated into a multi-user data repository operated through Opal-DataSHIELD, an interoperable open-source server application. Federated data analyses, without sharing or disclosing any individual-level data, is being performed to resolve various research questions emerging from dealing with the COVID-19 pandemic. Institute for Anthropological Research, Zagreb, Croatia, is one of the partners on the unCoVer project, together with our related third-party – Dubrava University Hospital, Zagreb, Croatia, providing clinical data on COVID-19 patients. Special emphasis will be given to our specific experience and lessons learned from participating in this project (data acquisition, ethical and GDPR issues, data analyses, influence of restrictions related to COVID-19 pandemic).

Presentation number: ANTR 5

DOMESTICATION OF CATTLE: CURRENT STATUS AND ANCIENT DNA PERSPECTIVES

Vlatka Čubrić-Čurik

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The domestication of animals is considered one of the most influential processes that have shaped the development and growth of human civilization. Cattle, which are divided into Taurus cattle (Bos taurus taurus) and Zebu cattle (Bos taurus indicus), are capable of converting large quantities of roughage into high-quality food, and the other benefits are also considerable. Therefore, cattle are often considered one of the most important domestic animals. Here, I first provide a historical overview of bovine domestication from the perspective of mitogenomes, Y chromosomes, and nuclear genomic markers. Second, I present the results of two recent genomic analyses of ancient cattle. Third, I show the results of our large-scale meta-analyses of the complete bovine mitogenome. Next, I present the archaeogenetics laboratory established this summer at the University of Zagreb - Faculty of Zagreb. Finally, I present our future perspectives in research on ancient genetics/genomics and domestication of animals.

Presentation number: ANTR 6

RUNS OF HOMOZYGOSITY AS AN IMPORTAN CONCEPT IN POPULATION GENOMICS

Ino Čurik

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Runs of homozygosity (ROH) are contiguous long stretches of homozygous genomic regions used to estimate realised autozygosity from high-throughput marker information. I will explain how ROH can be used to infer a number of important genomic population parameters. For example, ROH are used to estimate recent and distant inbreeding at the individual and population level. In addition, ROH are used to estimate inbreeding depression and to map genomic regions with detrimental effects. Genomic regions with above average occurrence of ROH or extreme ROH islands are used to identify genomic regions that exhibit patterns of positive selection. Finally, I will explain how ROH are used to estimate current and historical effective population size. All applications and examples presented are related to animals and humans.

Presentation number: ANTR 7

IMPLICATING EFFECTOR GENES AT COVID-19 GWAS LOCI IN DISEASE-RELEVANT IMMUNE CELL TYPES

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SARS-CoV-2 infection results in a broad spectrum of COVID-19 disease, from mild or no symptoms to hospitalization and death. COVID-19 disease severity has been associated with some pre-existing conditions and the magnitude of the adaptive immune response to SARS-CoV-2, and a recent genome-wide association study (GWAS) of the risk of critical illness revealed a significant genetic component. To gain insight into how human genetic variation attenuates or exacerbates disease following SARS-CoV-2 infection, we implicated putatively functional COVID risk variants in the cis-regulatory landscapes of human immune cell types with established roles in disease severity and used high-resolution chromatin conformation capture to map these disease-associated elements to their effector genes. This functional genomic approach implicates 16 genes involved in viral replication, the interferon response, and inflammation. Several of these genes (PAXBP1, IFNAR2, OAS1, OAS3, TNFAIP8L1, GART) were differentially expressed in immune cells from patients with severe vs. moderate COVID-19 disease, and we demonstrate a previously unappreciated role for GART in T cell-dependent antibody-producing B cell differentiation in a human tonsillar organoid model. This study offers immunogenetic insight into the basis of COVID-19 disease severity and implicates new targets for therapeutics that limit SARS-CoV-2 infection and its resultant life-threatening inflammation.

Presentation number: ANTR 8

GENDER DIFFERENCES AND GENETIC GEOGRAPHICAL VARIATION IN THE MEDITERRANEAN AREA OF COVID-19 DISEASE IN RELATION WITH INFERTILITY – OUTPUT OF EUROPEAN MEDIGENE PROGRAM

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Human populations were faced to COVID-19 pandemic due to emerging SARS-CoV-2 coronavirus from Wuhan (China) and with dramatic Public Health consequences. Although scientific community demonstrated an incredible innovation potential producing in one-year efficient vaccines, researchers are intrigued about ethnic and geographic diversity of endocrine or metabolic manifestations of COVID-19, including gender differences and effects on fertility. Man displays 1.5- and 2.5-fold higher Covid-19 mortality and SARS-CoV-2 infection, partially explained by differential expression of ACE2 (angiotensin-converting enzyme 2) receptor and TMPRSS2 (serine protease transmembrane protease serine2) located on Chr21q22.3. ACE2 is located on Chr-X, males being hemizygous while females expressing a mosaic pattern and potentially heterozygous with lower sensitivity to infection. Sex hormones regulate ACE2 expression and activity shifting the activity of Renin Angiotensin Aldosterone System (RAAS). TMPRSS2 gene has no sex-dimorphism but contains an androgen-responsive enhancer located on specific haplotypes more frequent in Europeans (e.g. Italians) and totally absent in East Asians. Gender disparity for Covid-19 infection also concerns the immune response, females being in general protected. Androgens have deleterious effects on thrombo-inflammation, raising the hypothesis on potential role of hyperandrogenism in women with polycystic ovary syndrome (PCOS) and infertility. In PCOS, there is indeed a 50% higher risk of Covid-19 infection, but vulnerability of patients may be explained by multiple factors such as chronic pro-inflammatory state (increase in TNF and IL-6), ethnic background (e.g. expression of Androgen Receptors in Black, Asian and Minority Ethnic groups) or low vitamin D levels. Recent GWAS studies indicated several susceptibility loci for Covid-19 infection with high ethnic disparities and structurally related to ancestral components of the human genome. All these epidemiological variations prompt scientists to further investigate susceptibility genes and long-term complications of Covid-19 infection.

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SARS-COV-2 AMONG SARAJEVO INHABITANTS - THE PRE-VACCINATION PERIOD

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Severe acute respiratory syndrome coronavirus 2 (SARS-CoV-2) is causing a novel COVID-19 infection. While the standard diagnostic test is molecular analysis using qRT-PCR, serological testing for COVID-19 is becoming increasingly important in research community and immunity surveillance efforts. Serological testing is performed on whole blood, serum, or plasma samples in a relatively simple and rapid procedure, requiring less expertise and simpler laboratory settings compared to molecular methods. Tests are designed to detect either total immunoglobulins (Ig) or to differentiate between immunoglobulins G (IgG) and immunoglobulins M (IgM) fractions. Preliminary study completed in 2020 offered the very first report on serological testing for SARS-CoV-2 in Bosnia and Herzegovina. Compared to the period April–July 2020, when anti-SARS-CoV-2 antibodies were detected in 3.77% of samples, one year later (May 2021) the estimated percentage within the same population of the urban Canton Sarajevo was 29.9% (5,406/18,066). Of all anti-SARS-CoV-2 Ig-positive individuals, 53.27% were men, and 69.00% were of 50 years of age or younger. Also, the current update found the individuals 50 years of age or younger to be more frequently anti-SARS-CoV-2 Ig positive compared to older individuals. On the other hand, higher median anti-SARS-CoV-2 Ig levels were found in individuals > 50 years old than in younger individuals, as well as in men compared to women. Seropositivity gradually increased from September 2020 to May 2021, with the lowest frequency of positive cases (3.5%) observed in September 2020, and the highest frequency (77.7%) in January 2021. Our results published within two separated publications in 2020 and 2022, provided important seroprevalence data that could help in planning restrictive local public health measures to protect the population of Sarajevo Canton, especially considering that at the time of the study the vaccines were virtually inaccessible to the general population not belonging to any of the high-priority groups for vaccination.

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ANTHROPOLOGICAL RESEARCH ON CROATIAN ISLANDS – CRIBS BIRTH COHORT

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The "Croatian Islands Birth Cohort Study (CRIBS)" is the first birth cohort study ever conducted in Croatia, designed to prospectively follow a sample of 500 pregnant women and their children up to two years of age in populations from Croatian Dalmatian islands (Hvar and Brač) and mainland population (city of Split with its surroundings). Data collected within the CRIBS cohort include data on social factors (age, marital status, education, employment, income, religious views, number of children), psychosocial factors (quality of life, health beliefs and attitudes, postnatal depression), lifestyle (smoking, alcohol consumption, dietary habits, physical activity), medical data (maternal obstetric data and medical history, potential pre-, peri- and post-natal complications, information on child's growth and development, feeding habits, allergies, vaccination, other medical issues etc.) and biological samples (blood samples from the mother and cord blood samples from the newborns). The results of the CRIBS study already showed that certain environmental and biological variables (e.g., maternal age, smoking, parity, lipid profile and fasting blood glucose level) are risk factors for the metabolic syndrome (MetS) and adverse pregnancy outcomes in this Croatian birth cohort. Especially pre-pregnancy BMI has been observed as an indicator of adverse maternal health during pregnancy (deviated biochemical parameters, high blood pressure), as well as an indicator of negative pregnancy outcomes. The advantage of the CRIBS cohort is the wealth of collected data (including biological samples of mother-child dyads), enabling future investigation of biological and environmental risk factors for the development of MetS and associated complex diseases.

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MEDITERRANEAN DIET ADHERENCE AND ITS ASSOCIATION WITH BIOLOGICAL MARKERS AND HEALTH IN DALMATIA

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Although the Mediterranean diet has beneficial effects on health, recent studies have shown low adherence in Europe. Rapid changes in the traditional way of life and the "westernization" of the diet in Mediterranean populations, especially in younger generations, has led to progressive abandonment of healthy dietary patterns. Adherence to the Mediterranean diet in Dalmatia, Croatia was assessed through the Mediterranean Diet Serving Score (MDSS) (Monteagudo et al. 2015) in two Dalmatian populations – pregnant women from Split and islands Hvar and Brač, and the adult population from the island of Hvar. Adherence to the Mediterranean diet was low to moderate among pregnant women, with no significant mainland–island differences. The highest adherence was observed among wealthier women with generally healthier lifestyle choices. The most significant mainland–island differences were observed for maternal lifestyle and socioeconomic factors (income, education, physical activity). Namely, adverse socioeconomic and lifestyle conditions were more pronounced in the island population, which, together with the observed non-Mediterranean dietary pattern, calls for more effective pre- and perinatal intervention strategies. When compared to the adult population from the island of Hvar, the young, reproductively active generation (pregnant women) in Dalmatia, Croatia, although having a higher education and socioeconomic status, exhibits a more adverse eating behaviour (lower adherence to the Mediterranean diet) and lifestyle (excessive smoking in pregnancy) than the older population from the same region. MDSS scores across aggregated age groups in both cohorts also showed significant association with age, blood lipid levels and smoking frequency.

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THE GENETIC LEGACY OF ARCHAIC HOMININ ADMIXTURE

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Modern humans overlapped for most of their history with other hominin groups, and an enduring question is whether or not our ancestors admixed with these, now extinct, hominin groups. DNA retrieved from Neandertal and Denisovan fossils revealed that our ancestors did admix with archaic hominin contemporaries, and remnants of Neandertal and Denisovan genomes still survive in present-day individuals. While the landscape of Neandertal and Denisovan ancestry in present-day populations has been outlined, the identification of archaic variation inherited from other hominin groups, such as H. erectus and H. floresiensis, is hampered by the absence of archaic reference genomes retrieved from their fossils. To overcome this limitation, we developed a “fossil free” statistical framework to detect putative “unknown” archaic sequences without relying on archaic genomes. We applied this method to genomic data we have generated from a contemporary pygmy population living on Flores Island (Indonesia), near the same cave where remains of the enigmatic small-bodied hominin species, H. floresiensis, were found. These data provide new insights into the interaction of archaic hominins in Island Southeast Asia and shed new light on how the unique biogeographical setting of the Flores pygmy population shaped their history and mechanisms of evolutionary change.

Presentation number: ANTR 13

OBESITY IN CHILDHOOD RELATED TO MATERNAL FACTORS DURING PREGNANCY IN THE ECHO-FGS COHORT STUDY

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The prevalence of obesity in US children has more than tripled in the past 40 years; hence, it is critical to identify potentially modifiable factors that may mitigate the risk. To examine the association between maternal pre-pregnancy body mass index (BMI), gestational weight gain (GWG) and child adiposity as measured by BMI, waist circumference and percent body fat in a racial-ethnically diverse cohort. In a prospective cohort study of healthy women without chronic disease, we examined the association between pre-pregnancy BMI, GWG and child adiposity. Children ages 4–8 years (n = 816) in the Environmental Influences on Child Health. Outcomes-NICHD Fetal Growth Studies were assessed. Trained study staff ascertained maternal pre-pregnancy BMI, GWG and child adiposity. The odds of child obesity (≥95th BMI percentile) increased independently for each unit increase in maternal pre-pregnancy BMI [OR = 1.12 (95% CI: 1.08, 1.17)] and for each 5-kg increase in GWG [OR = 1.25 (95% CI: 1.07, 1.47)]. The odds of child waist circumference (≥85th percentile) also increased independently for pre-pregnancy BMI [OR = 1.09 (95% CI: 1.05, 1.12)] and GWG [OR = 1.18 (95% CI: 1.04, 1.34)]. Maternal pre-pregnancy BMI and GWG were each independently and positively associated with child obesity and high child waist circumference.

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REGIONAL SENSITIVITY BAROMETER: STUDYING THE EFFECTS OF THE PANDEMIC ON DEVELOPMENT

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The regional sensitivity barometer evaluates the regional sensitivity of European regions and the regional needs amid the COVID-19 health and financial crisis. In this paper qualitative and quantitative indicators were combined in European level and then an overall sensitivity index was calculated for each average European region. Demographic, economic, health, tourism-related and COVID-19 pandemic-related data are taken into account, using differentiated weighting factors. The statistical data used was lastly evaluated on December 31st, 2020 in Eurostat database and processed in order to create the regional sensitivity for every country. The total index highlighted the final ranking between the European regions and formed the basis for the depiction of sensitivity on the European map. A series of conclusions came up; namely the existence of three groups of European Union (EU) member states in terms of regional resilience amid the COVID-19 crisis, as well as the extreme sensitivity of regions that later faced significant difficulty in handing the health and financial crisis, such as Portugal, Greece, Italy and the United Kingdom. In addition, Slovak Regions have shown preparedness to health threats. Comparing barometer results becomes clear that the sensitivity of a region is inextricably linked to its course over time, the chronic systemic pathogens and the readiness that has already developed in emergencies.