

present the Submitting Nodes. RDDR Network comprises clinicians from various dysmorphology centres in Lazio region with varying levels of expertise. Submitting Nodes transmit patients' clinical information and photographs. Accepted cases, appropriate for RDDR, are reviewed by RDDR dysmorphology experts throughout a forum section. Summary clinical reports are prepared an average a week after acceptance, depending on the timing and number of reviews, and are sent to the Submitting Nodes, and includes diagnostic suggestions and recommendations for further investigations or management of the patient.

Conclusions: RDDR Network enables clinicians of the submitting cases to access to a range of expert opinions and management advice, to increase individual and collective knowledge about dysmorphic rare conditions, and to raise current standards for the diagnosis, management, and increasing neonatologist skills in recognizing rare dysmorphic syndromes. RDDR was developed based on the experience of related European Dyscerne, A European Network of Centres of Expertise for Dysmorphology.

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P18.27

The psychosocial impact of risk-management for hereditary diffuse gastric cancer

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Between 30%-50% of cases of Hereditary Diffuse Gastric Cancer (HDGC) are caused by mutations in the E-cadherin gene. *CDH1* mutation carriers have an earlier than average age of disease onset, and greatly increased risks of developing cancer. Individuals identified as at-risk of HDGC, either because of their family history or as a result of DNA testing, need to make decisions about risk management, whether they will have risk-reducing surgery (total gastrectomy) or continue screening. This retrospective qualitative interview project was undertaken to explore i) decisions about risk management, ii) the psychosocial implications of undergoing risk-reducing gastrectomy (RRG), and iii) individuals' information and support needs. The investigation included 42 patients; 27 had undergone RRG and 15 were having endoscopic screening. Factors influencing surgical decisions include: experiences of disease, RR surgery and screening within the family, risk perception, social factors (e.g. familial obligations, life course issues) and tolerance of screening. Surgery was reported as having a number of psychological (e.g. impact on body image), social (e.g. impact upon family and earning potential) and physical costs and benefits. A number of information (e.g. impact on social life and reproductive options) and support (e.g. around eating practices) needs were identified.

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Approaching education in the genomic age - widest possible audience or narrow focus

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Education has been proposed for some time as the solution to managing the impact of emerging genetic and genomic technologies. Programs targeting the widest possible audience including the general public, individuals and families affected by genetic conditions, those at risk, schools and the professionals who care for them have been conducted with success by the Centre for Genetics Education NSW Health (CGE) in Australia for over 20 years. However health professionals report great concern for the lack of preparedness for new technologies. Given the enormity and rapid increase in patient and support information now available online, difficulties associated with measuring the impact of community awareness campaigns on health outcomes, the density of information for health professionals in this changing field and the limited resources allocated to education, the best way forward has recently been unclear. In addition approaches to education have significantly changed with the accessibility and popularity of social media, online programs and tools. Following a review in 2012, CGE has adopted a new model to meet this challenge narrowing its focus in order to deliver wider benefits. The approach targets non-genetic trained health professionals to develop skills and knowledge to manage the impact of genetic and geno-

mic technologies and in doing so strengthening and delivering wide patient benefits through their clinical practice including informed decision making, risk assessment, early appropriate referral, testing, detection, prevention, treatment and support. The model and strategies in place for 2013- 2015 are presented.

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Identification of a Single-Nucleotide Polymorphism of TAS2R38 gene (Bitter receptor gene). Genotype and phenotype characterization of a sample of Ligurian students

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In Liguria region an educational project for teachers and students of last two years of high school has been going on in collaboration with University of Genoa since many years involving lab activities, lectures and training sessions. During the last two meetings of Italian Society of Human Genetics a special session was dedicated to school to disseminate in Italy the Genoa experience. One of the most successful activities was the study of bitter-tasting ability by the analysis of the TAS2R38 gene polymorphisms, a didactic model of genotype-phenotype correlation. Three SNP of this gene - that codes for a transmembrane receptor - cause three amino acids substitutions in P49A, A262V, I296V, resulting in two common haplotypes, PAV (taster) and AVI (non taster). Individuals who have two dominant alleles (PAV/PAV or PAV/AVI) are sensitive to bitter substances, while those not sensitive (AVI/AVI) are recessive for this trait. The project involves thirty teachers and six hundred students and comprises a first part developed in schools: DNA extraction by mucose buccal cells, Bitter Taste Test by PTC paper, Questionnaire of food preferences, Bioinformatics. The second part takes place in a didactic laboratory of University: amplification of DNA trait including the SNP rs 713598, polymorphism identification by enzyme restriction and gel electrophoresis.

The activity allowed students:

- to evaluate genotype/phenotype correlation
- to calculate the frequencies of PAV and AVI genotypes
- to know that a number of SNPs are inherited as a haplotype
- to consider the role of genetic variability of taste perception

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The Clinical Laboratory Genetics profession in Portugal

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Portugal has several public and private laboratories dedicated to human genetic diagnosis, following high quality standards and professionals with qualifications in accordance with the standards of other European countries. The Clinical Laboratory Genetics profession is included in the career designated *Técnico Superior de Saúde, ramo de Genética (TSS-genética)*. To integrate this career a biomedical sciences background is needed (e.g. degree in Biology, Biochemistry, Pharmaceutical Sciences) followed by a 3 year internship that takes place in authorized genetic laboratories. The Portuguese health system is facing significant challenges due to reforms that are under implementation. Alongside the rationalization of expenses, it is very important to regulate the creation of several professions, an issue that has to be addressed by the Portuguese health services authorities.

The Workgroup for the Clinical Laboratory Genetics Specialty (GT-EGCL) designated by the Board of the Portuguese Society of Human Genetics (SPGH) has created a database/registry of the Clinical Laboratory Genetics Specialists working in public and private areas in Portugal. The results obtained by the GT-EGCL that will be presented, allowed the characterization of the Clinical Laboratory Genetics Profession in Portugal. The GT-EGCL fully endorses the inclusion of the Clinical Laboratory Genetics into Directive 2005/EC/36, towards the recognition of this specialty at the European level. This Europe-wide recognition is of tremendous importance not only to maintain the high-standards in clinical practice in human and medical genetics areas, but also for the cross-border mobility/recognition of these Portuguese professionals.

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