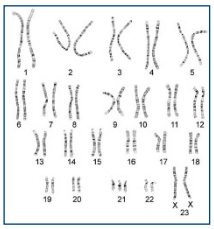
# Highlights of This Issue

### An ELSI Buffet

This special issue of *Genetics in Medicine* is devoted to articles that address ethical, legal, and social issues (ELSI) in genetics, coinciding with the 2011 ELSI Congress, to be held in Chapel Hill, North Carolina, April 12-14. The topics addressed this month highlight current dilemmas as researchers, practitioners, patients and the public all scramble to cope with rapidly evolving technological changes in the field. While these are a diverse set of articles, they are united by several common themes. Some provide information and guidance about complex issues in genetic testing regarding autism spectrum disorder (Miles; page 278), recommendations regarding informed consent for testing when non-genetic testing may lead to a genetic diagnosis (Chubak and colleagues; page 356), and difficulties involved in family communication regarding uninformative *BRCA1/2* results (Vos and colleagues; page 333). Others offer information that challenges accepted wisdom about the impact of genetic risk information. Fanos and colleagues (page 342) report on the relatively positive adaptation of a group of patients who opted for presymptomatic testing of familial ALS. Collins and colleagues (page 273) provide a systematic assessment of the impact of personalized genetic risk information on fatalism, and challenge the strength of evidence for such a view.

Other articles question the potentially harmful effects of new technology. Hawkins argues that gene patents in the UK have not limited access to genetic testing but observes this is due to the



patent system being ignored and warns against complacency (page 320). Hock and colleagues (page 325) find that genetic counselors have limited experience with direct to consumer (DTC) genetic testing while Wright et al (page 295) offer five requirements that would adequately protect the consumer when they engage in DTC genetic testing. Two articles address the changing moral focus of newborn screening, one featuring the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children Response to the President's Council on Bioethics report (page 301); and the other, an article by Richer and colleagues (page 305), reports on Canadian geneticists' views regarding the use of leftover bloodspots for medical research, which supports public engagement regarding both risks and benefits as policy is developed.

Lastly, two articles focus in different ways on concerns about social harms and justice in genetic testing and research. Levy and colleagues (page 349) present data from the first national sample of patients with newly diagnosed breast cancer at high risk for *BRCA1/2* mutations, finding low genetic assessment in general and striking underutilization of *BRCA1/2* testing among blacks and Hispanics. Rachul and colleagues; page 314) offer a linguistic analysis of racial terminology in genetics research, tracing population terminology from peer-review articles, to press releases, to newspaper articles to determine changes in language and frequency. Results indicated a wide variation in the frequency and terminology of population descriptor language used by genetic researchers and the media. One step to improve science communication policy, and combat inaccurate biological views of race, is surely more consistent and accurate use of population terminology.

# News Briefs

### Fetal DNA in the maternal circulation – real progress?

The well-documented presence of fetal DNA in the maternal circulation has long been a tantalizing target for the development of noninvasive prenatal diagnostic strategies. However, as attractive as this general idea has been the analysis of fetal DNA has never yet lived up to its promise. A report in *Nature Medicine* (published online 6th March 2011) may indicate that the tide is turning with regard to fetal specific DNA analysis for aneuploidy.

Papageorgiou et al, took an innovative approach to the analysis of fetal DNA in order to detect Down syndrome in a small sample of pregnant women by enriching for fetal specific methylated DNA and real-time quantitative PCR to achieve fetal chromosome dosage assessment in a noninvasive manner. In this small trial their approach provided correct diagnosis for 14 trisomy 21 (and 26 normal) cases. This is clearly a preliminary study having been implemented in a highly controlled context in a small sample of individuals. If however its promise is realized the noninvasive prenatal diagnosis of aneuploidies could finally be a reality – along with the inevitable controversy regarding techniques that make prenatal diagnosis much more routine.

### Highlights of the *AJHG*

### Another gene for MLC

This month's *AJHG* features an article by López-Hernández et al, describing the identification of a second gene, which when mutated, is responsible for megalencephalic leukoencephalopathy with subcortical cysts (MLC). The authors employed a proteomic based method to look for binding partners of the protein encoded by *MLC1*; mutations in *MLC1* cause the majority of cases of MLC. They discovered that an IgG-like cell adhesion molecule, GlialCAM, is a direct MLC1-binding partner. GlialCAM, also known as HepaCAM and encoded by the HEPACAM gene, was subjected to mutational analysis in 40 MLC patients without *MLC1* mutations and multiple different mutations were discovered. Mutant GlialCAM disrupts the localization of MLC1-GlialCAM complexes in astrocytic junctions in a manner reflecting the mode of inheritance. Thus, it appears that GlialCAM is required for proper localization of MLC1 and is the second gene found to be mutated in MLC.