GUIDANCE NOTES FOR:

(A) ADDITIONAL MANUSCRIPT FORMATS IN IJMEG

(B) SPECIFIC INFORMATION REQUIREMENTS FOR GENETIC ASSOCIATION REPORTS AND GENE META-ANALYSIS STUDIES

(C) GUIDANCE FOR THE DESCRIPTION OF MICRORNAS IN MANUSCRIPTS

(A) Additional IJMEG Manuscript formats - these other formats have some specific requirements:

- Technical or Methodological overviews
- Bioinformatics Application Note
- Commentaries
- IJMEG Editorials
- IJMEG Letters to the Editor
- Book Reviews
- Meeting Reports
- Meeting Announcements

General Principles

All manuscripts should be submitted according to IJMEG formatting policy.

Technical or Methodological overviews

These articles can follow the format and layout of the traditional reviews according to IJMEG formatting policy. The articles are expected to be instructional state of the art critiques on the practicalities and considerations of using particular techniques in experimental contexts. These articles should include brief individual introductions to methods being critiqued including discussion of the underlying assumptions and hypotheses of the techniques as well as outlining the practical and analytical pros and cons of each method. While solely theoretical discussions are accepted, the inclusion of illustrative sample data examples is also encouraged. These articles are expected to be between 4000-5000 words, excluding figures, tables, legends and references (not exceeding 100) are no word or reference limits but will be peer reviewed in a similar way to reviews for factual accuracy and pre-publication proofs will be made available to authors for comment.

Bioinformatics Application Note

These articles do not necessarily follow the traditional format and unlike a number of the other articles we do not wish to be overly prescriptive in terms of structure. However this format shares the need of other formats of having an Abstract, which is important for PubMed searchers. In addition to this suggested headings for this format would be: (A) Motivation/Background of need, (B) Implementation/Methodological Approach – which should where possible include relevant code, (C) Validation – which should include demonstrable examples of its functionality as much as possible. This format should also include the usual Acknowledgements and appropriate referencing according to the IJMEG style (Please download EndNote X Style or Reference Output Style for Reference Manager: IJMEG_RefManfor)

Commentaries

The main objective of commentaries is to express personal opinions on a topical issue within any area relevant to the study of the molecular basis of disease. This could include viewpoints on experimental or analytical approaches that are being used or current assumptions and conclusions being presented in any context. Articles of this nature can be provocative, within reason, and may encourage further debate on a topic, however, if applicable authors will be offered and expected to contribute responses to any submissions to IJMEG arising from their commentary. This article format should be no longer than 2000 words and can include up to two tables or figures and no more than 25 references. Proofs will be made available to authors for comment.

IJMEG Editorials

Editorials in International Journal of Molecular Epidemiology and Genetics (IJMEG) will normally be provided by the Editors-in-Chief or be solicited by them. These will be subject to a review process. Editorials should not be longer than 1000 words and can include a single table or illustration (but not both). There should normally not be more than 10 references. Proofs will be made available to authors for comment.

IJMEG Letters to the Editor

This format allows for the submission of responses to articles either published in IJMEG or elsewhere. Such articles will not be simply an expression of personal viewpoint, such as is captured by Commentary articles and instead will be expected to include data that form basis of and are the motivation behind their response. Letters to the Editor will be subject to peer review. Contributors of such formats should commence their submissions with immediate reference to the originating article they are commenting on and use appropriate referencing. Letters ordinarily should not be longer than 1250 words (excluding legends and references) and should not contain more than 10 references. Letters can present new and relevant data in up to two illustrations (i.e. tables or figures. IJMEG reserves the right to publish letters in a shortened format at the discretion of the editors or if deemed appropriate to request the submission in an alternative more appropriate format. IJMEG also reserves the right to invite the authors of the originating article to provide additional comments or rebuttals where appropriate. Proofs will not be sent to the authors.

Book Reviews

IJMEG welcomes book publishers to submit books or initially a list of possible book titles with a view to their review in IJMEG, for which IJMEG will facilitate the identification of reviewers. In such instances where a book review is completed and published in IJMEG, the publisher, not the author(s), will be liable to meet the associated publishing charges. The journal will also consider unsolicited book reviews from authors but subject to there being no affiliation, commercial interest or otherwise with the book being reviewed and a statement of declaration to this effect must be made with the cover letter. For such submissions authors are initially encouraged to contact editorial@ijmeg.org to discuss their intention and the author will be responsible for meeting the associated publishing costs. All Book Review submissions should be no longer than 600 words, including details of the author(s) or editor(s), title, date, publishers, number of pages, cost, ISBN of the book being reviewed, as well as any additional references as well as the reviewer(')s name(s), institution and contact email address details.

All Book Reviews will subject to editing if necessary by the IJMEG Editorial Office to fit with IJMEG's format and style which also aims for each review to ensure appropriate grammar is used, to reduced wordage where appropriate and if applicable to present the article in a manner most appropriate to the broad readership of IJMEG. Reviewers will be given an opportunity to accept or reject the suggestions made to the articles by the IJMEG Editorial Office. Once authors have been responded the Editorial office suggestions, this will be considered the final edit (proofs will not be sent back to the authors) and taken by the IJMEG copy editor for inclusion in the next issue of IJMEG.

Meeting Reports

This offers a flexible format but the title of the article should include details of the title of the event, the dates on which it was held and the meeting location. This should then be followed by details of author(s), their affiliations and email addresses. There is no word limit for this format of article but is subject to standard IJMEG publishing charges. Any affiliation, involvement or lack thereof, of the author(s) with the organisation of the event or any commercial interests arising from or connect to the event should be stated explicitly at the end of the article under a heading entitled Declaration after any Acknowledgements.

Meeting Announcements

IJMEG will consider providing links to or the online publication of either a textual description or the formal marketing literature of a forthcoming event. All enquiries on this matter should be directed to the editor@ijmeg.org in the first instance.

(B) Genetic Association Reports and Gene Meta-Analysis Studies - Specific Information Requirements

Articles submitted for consideration as a Genetic Association Reports are intended to be brief and to the point. The scope of these articles can be studies of individual or multiple genes right up to the level of Genome Wide Association Studies (GWAS) and can involve single or multiple sites of variation. IJMEG will also consider studies of genetic linkage.

To help standardise the presentation of these studies and promote the greater ease of reading and replication efforts the following additional requirements should be incorporated in articles in addition to the standard IJMEG Author Guidelines. (To facilitate the meeting of these guidelines this has been designed for print off and can serve as a checklist).

Title

☐ Diseases or phenotypes the report refers to must be included in the title.

Introduction.

☐ Any diseases referred to should provide a relevant Online Mendelian Inheritance in Man (OMIM) number;

- avoid exhaustive background information, focus on pathogenic pathways of relevance to the gene(s) of study.

Materials & Methods.

- Every effort should be made (through collaborative efforts if necessary) to investigate as sizeable a dataset as possible;
- Explicitly comment on the statistical power of the study;
- Any follow-up or replication studies where data from subsets of the cases and controls have already been published on the same gene and variations require these studies to be cited;
- In the case of age-related diseases mean ages of onset (and age at death if applicable) for cases (age at collection for controls) should be provided and if relevant the percentage of subjects the mean values are based upon;
- Any data or information extracted from *www* hosted databases e.g. www.hapmap.org should be referred to according to guidelines of the source website and if not already stated the date of access and any appropriate build or version numbers;
- Where appropriate all genes should be referred by their approved gene symbol in accordance with recommendations made by the **HUGO Gene Nomenclature Committee**;
- Reference to Single Nucleotide Polymorphisms (SNPs) should where possible refer to appropriate reference SNP (rs) numbers. In the event that there is not an rs number available for variant(s) that are described, for the purposes of the manuscript either use abbreviations by which they have been previously referred or for novel variants create abbreviations that are based upon the gene symbol and include the genomic location of the variant(s) relative to their human chromosomal reference sequence (noting any sequence build numbers). Also state if rs numbers have been sought for novel variants. Equally, other non-SNP genetic variants discussed should, where possible, be described by officially recognised abbreviations or where not available based upon abbreviations by which they have been previously referred or for novel variants create abbreviations based upon the gene symbol and including information on the appropriate genomic location relative to the human chromosomal reference sequence (noting any sequence build numbers);
- Statistical methods should refer to the probability values (p-value) used to define statistical significance; to all forms of analysis used and the contexts in which they have been used and to state the form of analysis used to correct for multiple testing.

Results

- This section should be used only to present the logical progression of analysis followed in the study and the corresponding results in a factual manner;
- Any reports of diseases where there are already well accepted susceptibility genes should in addition to presenting raw data from the study in question, also incorporate or refer to stratified or regression analysis involving the appropriate "risk" data on these genes to demonstrate that consideration has been given to any potential interactions;

☐ Probability values (p-values) that have or have not been corrected for multiple testing should be prefixed respectively as *corr*-(i.e. corrected) or *un-corr* (i.e. uncorrected) where appropriate;

Discussion

☐ Avoid excessive duplication of background information already provided in the introduction;
☐ Data trends that do not reach statistical significance of association should not be overstated in the discussion;
☐ Any replications studies should be contextualised with respect to existing evidence at the time of submission, if available refer to relevant meta-analyses to minimise exhaustive referencing;
☐ Without over-speculating, authors should suggest (supported by published evidence where available) plausible biological mechanisms by which gene variants might contribute to changes in biological function that may contribute to, cause or attenuate the progression and course of disease, all of which will serve to direct future research.

(C) GUIDANCE FOR THE DESCRIPTION OF MICRORNAS IN MANUSCRIPTS

There is variability in the literature around the use of capitalisation when describing miRNAs and as a result there is ambiguity in the literature about what is being described e.g. hairpin RNA sequence vs genomic locus in various studies involving miRNAs.

In the interests of standardisation, the absence of any agreed conventions and following consultation with the hosts of miRBASE (http://www.mirbase.org/), IJMEG has introduced requirements for all submissions in which miRNAs are described to follow conventions set out below. As a general rule, manuscripts significantly benefit from explicit introduction and clarification of the nature of a particular entity (e.g. miRNA gene, precursor, mature form etc) and for this to be done consistently throughout the manuscript eg "the mature miR-100".

IJMEG conventions to be used include:

- When referring to miRNAs use the same ID format as that used in miRBASE (http://www.mirbase.org/).
- At the first mention of miRNAs in the text please hyperlink the miRNA symbol to its corresponding entry on miRBASE (http://www.mirbase.org/).
- For the description of gene loci describing miRNAs:

  For human related applications, by extension to RNAs, the use of capitalisation and italics for *GENE* names will follow. In other words the gene *symbol* for any miRNA should use the relevant symbol as is listed in miRBase (as is done by HGNC) and it should be italicised and capitalised when referring to the gene encoding it in manuscripts e.g. *MIR100*. For descriptions of animal miRNA genes the format *mir-100* should be used.
- For the description of miRNAs themselves:

  To differentiate between hairpin and mature miRNA sequence, the convention mir-100 should be used for hairpin sequence whereas miR-100 should be used to describe the mature sequence.