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### DESCRIPTION

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The *European Journal of Medical Genetics* (EJMG) is a peer-reviewed journal that publishes articles in English on various aspects of **human** and **medical genetics** and of the **genetics** of experimental models.

Original clinical and experimental research articles, short clinical reports, review articles and letters to the [editor](#) are welcome on topics such as :

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### AUDIENCE

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Researchers, Geneticists, Cytogeneticists.

### IMPACT FACTOR

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Submission

*Type of manuscript*

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Define any abbreviations that are not standard in this field in the text, by putting abbreviation between brackets after the full text (example: .... In autism spectrum defect (ASD)... Noonan syndrome (NS) ...). Ensure consistency of abbreviations throughout the article.

### *Acknowledgements*

Collate acknowledgements in a separate section at the end of the article before the references and do not, therefore, include them on the title page, as a footnote to the title or otherwise. List here those individuals who provided help during the research (e.g., providing language help, writing assistance or proof reading the article, etc.).

### *Math formulae*

Present simple formulae in the line of normal text where possible and use the solidus (/) instead of a horizontal line for small fractional terms, e.g., X/Y. In principle, variables are to be presented in italics. Powers of e are often more conveniently denoted by exp. Number consecutively any equations that have to be displayed separately from the text (if referred to explicitly in the text).

### Footnotes

Footnotes should be used sparingly. Number them consecutively throughout the article, using superscript Arabic numbers. Many wordprocessors build footnotes into the text, and this feature may be used. Should this not be the case, indicate the position of footnotes in the text and present the footnotes themselves separately at the end of the article. Do not include footnotes in the Reference list. *Table footnotes* Indicate each footnote in a table with a superscript lowercase letter.

### Specific templates for the main text section

**Although some manuscripts may escape the general recommendations that follow (after editor's acceptance), the manuscripts should follow the general templates presented below.**

#### Template for Clinical and Experimental research articles

**Introduction.** This section contains general informations about the topics addressed by the research. If a general review of the literature is necessary (discouraged in *Short Reports*), it should be set in the *Introduction*, in order to separate it clearly from the *Discussion* itself. There is no subheadings in the introduction. **Patient Data / Material.** *Patient data* heading is used to describe the human subjects under study. It can be subdivided if necessary in several subheadings: ascertainment of patients, patient reports, family reports, patient 1, patient 2... The term "case report" is not accepted, and patient should not be identified by their initials or file number. Patients described in a manuscript should be regarded with sensitivity. Stigmatizing terms should be avoided. As often as possible, refer to pictures with relevant features. Facial dysmorphism is better illustrated by a picture than by an extensive description. For large series of patients, use Tables instead of text as often as possible. Keep only the most pertinent points in the main text, and avoid duplication in the main text of informations that are shown in the table.

*Material* replaces the *Patient Data* section when the research is not focalized on direct observation human beings, but (for instance) on the analysis of large populations with no description of individuals (such as studies based on cohorts or registries), or experimental models **Methods.** This section describes procedures (in conjunction with appropriate references) in order to allow readers to understand how the experiments were performed, with sufficient details to allow all procedures to be repeated. This heading can be further subdivided according to manuscript specificities. Statistical methods deserve a specific paragraph, or a subheading. Widely used methods (such as DNA extraction, classic Sanger sequencing, conventional karyotyping or FISH techniques...) do not have to be described, except for the aspects that are specific to the addressed problem (for instance : type of an array, software parameters used for analysis, references of BACs used in a FISH experiment...). **Results.** The section may be divided with subheadings. As a rule, the results should not be commented or discussed in this section. **Discussion.** The discussion section should be focused on the discussion of the results, perspectives and hypotheses, and ends with a paragraph of conclusion. The section may be divided with subheadings. It should not be redundant with the Results section. Usually, a general introductive review of should not be presented in this section, but rather in the introduction. **7.,...** **Facultative sections:** Acknowledgements, Accession numbers, etc.

#### Template for Clinical Reports and New clinically defined syndromes

**Introduction** (see Research article template) **Clinical report.** This heading is used to describe the human subjects under study and the results of the investigations. It can be subdivided if necessary in several subheadings: 2.1. Patient 1, 2.2. Patient 2... Please provide sex, ethnicity, parental age, biometry at birth and gestational age, biometry at last investigation -with centiles and/or standard deviations. Only relevant (normal and abnormal) results needs reporting. The can be presented in a paragraph at the end of the clinical report, or below a separate subheading when several patients have the same anomaly. Widely used methods (such as DNA extraction, classic Sanger sequencing, conventional karyotyping...) are not described, except for the aspects that are specific to the addressed problem (for instance: type of an array, software parameters used for analysis, references of BACs used in a FISH experiment...). **Discussion.** (see Research article template) - **5.,...** **Facultative sections:** Acknowledgements, Accession numbers, etc. **References** are limited to 25.

#### Template for Exome Report

**Abstract.** The abstract should include the list of the gene symbols that are discussed in the variation description section **Clinical description.** Please provide sex, ethnicity, parental age, biometry at birth (and gestational age), biometry at last investigation - both with centiles and/or standard deviations,

psychomotor development (if possible include IQ or DQ, with the test used and at what age), phenotype at last examination, current age. Use the Standard Terminology proposed Elements of Morphology (vide supra). Give pertinent family information **Methods**.

Capture method Type of sequencer Variant analyse strategies (home-made pipeline, public tools...)

General statistics on sequencing quality, in a Table format (only analyse variants of less than 3% incidence) List of database checked (1000 Genomes,...) and date of check **NGS summary for each sample**

9081\_ejmg\_gfa\_exome report template1.jpg **Variations of interest**. All variants presented in this section have predicted pathogenicity by at least one prediction program and occur in genes could be hypothesized to be associated with the phenotype based on current knowledge of gene function, pathway, expression pattern, etc... All variants must have been checked by Sanger sequencing in the trio. Other variants will only be available online (see Supplementary data). Secondary findings unrelated to the phenotype will not be reported.

Sequence variation(s) reported in the main section should be ordered similarly

heterozygous variants/indel present in the proband and absent in parents and controls; homozygous variants/indel and hemizygous X variants never reported in controls; and genes with two rare non synonymous variants/indel with mean allelic frequency <0.03, present in the proband, but not seen together in the parents and controls.

Limit reports in this section to (1) indels with >5 variant reads and a variant/reference read ratio >0.3 and (2) single nucleotide variations that that have coverage  $\geq 10$  in proband and parents, whose raw alignment has been checked in parents, Mandatory information are listed below. If more than one significant variation is identified, results are presented in a tabular form using this template, direct or transposed (an exemple of exonic, intronic and INDEL is given).

9081\_ejmg\_gfa\_exome report template2.jpg

The full list of variants is added as supplementary material with similar dataset **Discussion**. Argue the selection of candidate genes that you kept in this section. **Facultative sections**: Acknowledgements,...etc. **Supplementary Table (mandatory)**. Include full data set in an Excel table, including variants for which Sanger sequencing was not performed, or not predicted to be pathogenic by prediction softwares. Requested information is similar to previous section. Include here at least: **References**. Maximum 2 references by candidate gene.

*Template for Array Report*

**Clinical description**. Please provide sex, ethnicity, parental age, biometry at birth (and gestational age), biometry at last investigation - both with centiles and/or standard deviations, psychomotor development (if possible include IQ or DQ, with the test used and at what age), phenotype at last examination, current age. **Methods**. For Array-CGH give precise type of the array, the software used for analysis... The draft version of the genome used for reporting the anomalies must be given when the position of the nucleotide are given. End the section with the method of confirmation: FISH, microsatellite, MAPH, MLPA... and the results of family screening. Confirm the status of the region in DGV and/or DECIPHER databases (or another similar reference database of human genomic variants).

**Genomic rearrangement**. For BACs, give the name and position of the probe (when interrupted), or the name and position of the 2 probes between which each chromosomal break occurred. For array technologies, give the positions in nt of the 2 flanking probes with the number of the genome draft used as reference. Precise minimal and maximal sizes and parental origin (if studied). **Discussion**. Do not repeat the phenotypic description but indicate most characteristic feature(s). Compare with previous report(s) on similar chromosomal imbalances... stressing on possible genotype/phenotype correlations. Candidate genes and may be discussed here. If necessary, the list of all genes present in the rearranged region can be added as a Supplementary Table. **Facultative sections**: Acknowledgements, Accession numbers, etc. **References**. Maximum 5 references.

### **Artwork**

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## Electronic artwork

### General points

- Make sure you use uniform lettering and sizing of your original artwork.
  - Preferred fonts: Arial (or Helvetica), Times New Roman (or Times), Symbol, Courier.
  - Number the illustrations according to their sequence in the text.
  - Use a logical naming convention for your artwork files.
  - Indicate per figure if it is a single, 1.5 or 2-column fitting image.
  - For Word submissions only, you may still provide figures and their captions, and tables within a single file at the revision stage.
  - Please note that individual figure files larger than 10 MB must be provided in separate source files.
- A detailed guide on electronic artwork is available on our website:

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**You are urged to visit this site; some excerpts from the detailed information are given here.**

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EPS (or PDF): Vector drawings. Embed the font or save the text as 'graphics'.

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TIFF (or JPG): Bitmapped line drawings: use a minimum of 1000 dpi.

TIFF (or JPG): Combinations bitmapped line/half-tone (color or grayscale): a minimum of 500 dpi is required.

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- Supply files that are too low in resolution.
- Submit graphics that are disproportionately large for the content.

### Figure captions

The digital image files are entered separately from the manuscript text file in the web site (do NOT include the images with the manuscript text file). Image sizes for review purposes should be sufficient to allow reviewers to clearly see the elements of the figure. Color images and figures are strongly encouraged, as they are published online at no cost to the authors. Color images are placed against a white background wherever possible. For each figure, an individual high resolution digital files must be uploaded.

For figures that have multiple panels, the labels should be set in uppercase letters in Arial font, taking the scale reduction in account for the font size. Do not include separate panels on multiple pages. White or black arrows can be added on the pics for sake of clarity. Micrographs should be provided with a scale bar, if appropriate. Magnification can be added in the caption. Pedigrees should be drawn according to the published standards of Am J Human Genetics 1995 56:745-752)

**Legend of figures.** A caption should comprise a brief title (**not** on the figure itself) and a description of the illustration. Legends are presented on a separate page of the main text file. The legend should explain what is being illustrated without the need to refer back to the text. Authors should describe the abnormal findings illustrated by the figure and not mention only (for instance) "abnormal aspect of the X rays of patient 1". White or black arrows can be added on the pics for sake of clarity. If references are embedded within a figure, they must also be cited within the figure legend. Keep text in the illustrations themselves to a minimum but explain all symbols and abbreviations used.

### Figure captions

Ensure that each illustration has a caption. A caption should comprise a brief title (**not** on the figure itself) and a description of the illustration. Keep text in the illustrations themselves to a minimum but explain all symbols and abbreviations used.

## Tables

Tables are self-explanatory and do not duplicate information present in the text. Each table must have a title. If necessary, place footnotes to tables below the table body and indicate them with superscript lowercase letters. Avoid vertical rules. Any symbols and abbreviations used in the table body must be defined in a footnote to the table. Each Table is presented on a separate page, followed by its legend

**Legend of Tables.** The legend should explain the Table without the need to refer back to the text.

## References

### *Citation in text*

Please ensure that every reference cited in the text is also present in the reference list (and vice versa). Any references cited in the abstract must be given in full. Unpublished results and personal communications are not recommended in the reference list, but may be mentioned in the text. If these references are included in the reference list they should follow the standard reference style of the journal and should include a substitution of the publication date with either 'Unpublished results' or 'Personal communication'. Citation of a reference as 'in press' implies that the item has been accepted for publication.

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2. Two authors: both authors' names and the year of publication;
3. Three or more authors: first author's name followed by "et al." and the year of publication.

Citations may be made directly. Groups of references should be listed first alphabetically, then chronologically.

*Examples:* "as demonstrated in wheat [Allan, 1996a, 1996b, 1999; Allan and Jones, 1995; Johnson et al., 1992].

Kramer et al. [2000] have recently shown ...."

*List of references:* References should be arranged first alphabetically and then further sorted chronologically if necessary. More than one reference from the same author(s) in the same year must be identified by the letters "a", "b", "c", etc., placed after the year of publication.

### *Examples:*

Reference to a journal publication:

Van der Geer J, Hanraads JAJ, Lupton RA. The art of writing a scientific article. *J Sci Commun* 2000;163:51-9.

Reference to a book:

Strunk Jr W, White EB. *The elements of style*. 3rd ed. New York: Macmillan, 1979.

Reference to a chapter in an edited book:

Mettam GR, Adams LB. How to prepare an electronic version of your article. In: Jones BS, Smith RZ, editors. *Introduction to the electronic age*. New York: E-Publishing Inc.; 1994. p. 281-304.

Note shortened form for last page number. e.g., 51-9, and that for more than 6 authors the first 6 should be listed followed by "et al."

For further details you are referred to "Uniform Requirements for Manuscripts submitted to Biomedical Journals" (J Am Med Assoc 1997;277:927-34), see also [http://www.nlm.nih.gov/tsd/serials/terms\\_cond.html](http://www.nlm.nih.gov/tsd/serials/terms_cond.html).

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