EUROPEAN JOURNAL OF MEDICAL GENETICS

AUTHOR INFORMATION PACK

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DESCRIPTION

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 :

- Dysmorphology and syndrome delineation
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Types of paper

Submission
Type of manuscript
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In clinical research articles, the experimental data are the support to clinical investigations.

See template below. **Experimental research**. Results of experimental investigations related to medical genetics, such as cell biology and animal models of human disorders, technology-oriented research, bioinformatics research... In experimental research articles, the experimental data are the most important elements of the work. See template below.**Genetic forum**. The manuscripts of this section may address the practice of medical genetics, and all its correlates: ethics, social issues, organization, perspectives... There is no specific format for Genetic forum articles.**Clinical Reports**. Report of significant new findings in a single patient, a single pedigree, or a small series of patients with a known disorder. Not including the title, list of authors, affiliations and figures, the manuscript must not exceed fifteen manuscript pages (600 lines) including legends, tables and a maximum of 25 references.**New clinically defined syndrome**. Clinical reports of "new" syndromes of unknown etiology. Reports will only be considered when at least results of molecular karyotyping (by CGH- or SNP-arrays) are provided. A specific template is found below.

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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.

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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 subdived 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 been 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 ≥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).

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The full list of variants is added as supplementary material with similar dataset **Discussion**. Argument 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 perforrmed, 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.

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