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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 :

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AUDIENCE

Researchers, Geneticists, Cytogeneticists.

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Descriptive terminology for dysmorphology

If the manuscript reports specific morphological features of human subjects or other developmental anomalies, authors should use the [Human Phenotype Ontology](#) (HPO) which provides a structured, comprehensive and well-defined set of descriptors. Alternatively, for craniofacial features, hands and/or feet, the authors may use the [Elements of Morphology: Standard Terminology](#) (Am J Med Genet 2009, 149A (1) : 1-127).

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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.).

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Template for the main section of Research Reports

Research manuscript must be kept concise, but there is no mandatory requirement for length or number of references.

Introduction: This section contains general information about the topics addressed by the research. If a general review of the literature is necessary, it should be set in the Introduction, 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 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 information that are shown in the table. **Material** section 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) 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 per manuscript specificities. Statistical methods deserve a specific paragraph, or a subheading. Widely used methods (such as DNA extraction, classic Sanger sequencing, SNP- or CGH-array karyotyping) do not have to be described, except for the aspects that are specific to the addressed problem (for instance: type of an array, software 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.

Template for Clinical Reports and Array reports

The main section of manuscript must not exceed fifteen manuscript pages (600 lines) and a maximum of 25 references.

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 (Family 1, Patient 1).

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 are not described, except for the aspects that are specific to the addressed problem (see Method in Research Reports).

In Array Reports, confirm the status of the region in the dedicated databases (DECIPHER, ClinVar or similar). If necessary, the list of all genes present in the rearranged region can be added as a Supplementary Table.

Discussion: (see Research report template) Do not repeat the phenotypic description but indicate most characteristic feature(s).

References: They are limited to 25.

Template for Exome Report

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Abstract: The abstract should include the list of the gene symbols that are discussed in the variation description section

Clinical report:(see Template for clinical 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, Use the Standard Terminology proposed Elements of Morphology (vide supra).

Methods:Describe capture method, type of sequencer, strategies for variant analysis (home-made pipeline, public tools). Give general statistics on sequencing quality (average depth of sequencing, % of captured regions with depth greater than 10). List the databases checked (ExAC) and date of check.

Results: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 The full list of variants and there is presented as supplementary material.

Discussion:(see Research report template).

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