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WG04-2012: SEQUENCE VARIANT DESCRIPTIONS

CHARTER

1. Name

1.1. The name of the Working Group shall be WG04-2012: SEQUENCE VARIANT DESCRIPTIONS

2. Purpose

2.1. The Working Group shall:

2.1.1. publish a review of current sequence variant description recommendations; and

2.1.2. prepare pursuant to PD06-2011: Standards Development Process an exposure draft of an HVP Standard or Guideline covering:

2.1.2.1. methods for the unambiguous description of all sequence variant at the DNA, RNA and protein level; and

2.1.2.2. is usable by both humans and computers.

2.1.3. provide advice to the Sponsoring Council about how the resultant HVP Standard or Guideline should be maintained into the future.

3. Operations

3.1. The Working Group will operate in accordance with the Human Variome Project's Standards Development Process (see *PD06-2012: Standards Development Process*).

3.2. The scope of work may be modified at any time by the Sponsoring Council. A request to modify the scope of work can be made in writing by the Chair of the Working Group to the Chair of the Sponsoring Council.

4. Sponsoring Council

4.1. The International Scientific Advisory Council is the Sponsoring Council for this Working Group.

5. Duration

5.1. The Working Group shall have two (2) years from the date of its establishment to prepare and submit an Exposure Draft of an HVP Standard or Guideline.

5.2. The Working Group may be disbanded before the completion of two (2) years, pursuant to *PD06-2012: Standards Development Process*.

6. Membership

6.1. Membership of the Working Group is at the discretion of the Sponsoring Council and/or the Chair of the Working Group.

6.2. The Chair of the Working Group is Johan den Dunnen (ddunnen@humgen.nl)

6.3. A quorum of 50% of Working Group members is required for all votes and meetings.

6.4. Decisions will be by consensus of the members present.

6.5. The Chair should make every effort to ensure that the following are represented on the Working Group:

6.5.1. Gene/Disease Specific Database Curators, including:

6.5.1.1. Raymond Dagleish; and

6.5.1.2. Peter Taschner.

6.5.2. The ISCN Committee;

6.5.3. Commercial diagnostic laboratories;

6.5.4. Central repositories;

6.5.5. The Cytogenetics community; and

6.5.6. Clinical geneticists.

6.6. Individuals who are not members of the Working Group may be invited to attend meetings at the request of the Chair to provide advice and assistance where necessary. They shall have no voting rights and may be requested to leave the meeting at any time by the Chair.

6.7. Members will cease to be a member of the Working Group if they:

6.7.1. resign from the Working Group; or

6.7.2. fail to attend two (2) consecutive meetings without providing apologies to the Chair.

7. Meetings

7.1. The Working Group shall set its own meeting schedule at the discretion of the Chair.