Argumentation for Scientific Claims in a Biomedical Research Article

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Abstract

This paper provides an analysis of some argumentation in a biomedical genetics research article as a step towards developing a corpus of articles annotated to support research on argumentation. We present a specification of several argumentation schemes and inter-argument relationships to be annotated.

1 Introduction

This paper provides an analysis of some argumentation in a biomedical genetics research article (Schrauwen et al., 2012), as a step towards developing a corpus of articles annotated to support research on argumentation (Green, 2014). For each argument for a scientific claim in an article, we would like to annotate its premises, conclusion, and argumentation scheme. In addition we would like to annotate certain relationships between pairs of the arguments, e.g., where one provides support for the premise of another. In order to develop an annotation system that can be consistently applied by different coders or by the same coder at different times, it is necessary to develop a precise specification of each argumentation scheme and inter-argument relationship. In this paper, we present a specification of several argumentation schemes and inter-argument relationships to be annotated.

The main claim of (Schrauwen et al., 2012), summarized in its title, is that a certain variant, c.637+1G>T of the *CABP2* gene, is a cause of moderate-to-severe, autosomal recessive non-syndromic hearing loss (arNSHL). According to our analysis, the argumentation in the article serves at least four types of discourse goals. The first is to persuade peer reviewers that the article is worthy of publication. The second is to persuade the audience that the scientific methodology used by the authors was sound and that the evidence so acquired is reliable. Arguments for the third type support or defend the scientific claims of the article. Arguments for the fourth type support the practice implications, i.e., the authors' suggested application of the scientific contribution to medical practice. The planned corpus will be annotated for arguments of the third and fourth type. In the next section, we briefly discuss the first two types, before focusing on the third and fourth types.

2 Discourse Goals

2.1 Novelty and Significance

The Knowledge Claim Discourse Model (KCDM) (Teufel, 2010) provides a multi-level description of consecutive text segments of a scientific article in terms of the "knowledge claims", or purported scientific contribution of the article. "The top level … formalizes the authors' high-level rhetorical goals, which serve to defend the new knowledge claim of an article against possibly hostile peer review … For instance, authors must argue that their new knowledge claim is novel and significant, and sufficiently different from already existing knowledge claims to warrant publication" (p. 102). According to Teufel, these arguments are not "directly textually expressed", but can be inferred by the reader from lower-level rhetorical moves that "often contain meta-discourse phrases such as 'In contrast to traditional approaches'.

In the Introduction section of (Schrauwen et al., 2012) the significance of the search for causes of arNSHL can be inferred from text such as "Hearing loss is a common sensory disorder that

can significantly impact quality of life" (p. 636). An argument for novelty is given in Excerpt 1.

Excerpt 1:
"Most families segregating arNSHL typically have a prelingual, bilateral, severe-to-profound hearing loss. An exception is found with mutations in *TECTA* … and *STRC* …; these mutation cause moderate-to-severe hearing loss … Recently, we identified a locus associated with arNSHL on 11q12.3-11q13.3 (DFNB 93) in an Iranian family that also presents a similar moderate-to-severe hearing loss phenotype … Here, we report that a mutation in *CABP2* … is the cause of DFNB93 moderate-to-severe hearing loss and reveal a role for CaBP2 in the mammalian auditory system." (p. 636)

By design, the KCDM does not address argumentation whose identification requires understanding of scientific content. Thus, the KCDM is not concerned with characterizing the other uses of argumentation that we found in the genetics article.

2.2 Methodological Soundness

The Results section of (Schrauwen et al. 2012) employs a narrative style reporting the sequence of events in the authors' scientific investigation, the reasons for the actions taken during the investigation, and the results of those actions. In so doing, the authors provide implicit arguments for the soundness of their scientific methodology. (The Materials and Methods section of the article provides more details about the methodology.) For example, Excerpt 2 provides a reason for the authors' decision to sequence a certain region of the genome of a certain individual (V:14).

Excerpt 2:
"The DFNB93 region contains more than 300 annotated and hypothetical genes, and several genes are expressed in the mouse and human inner ear. Because there are many strong candidates in the region, we sequenced all genes and noncoding genes in this region by using a custom DNA capture array to identify the disease-causing mutation in one affected individual from the family." (p. 639)

This passage can be analyzed as an instance of a type of Practical Reasoning argument whose discourse goal is to justify the authors' action (sequencing the DFNB93 region by using a custom DNA capture array) in order to achieve the authors' goal (to identify the disease causing mutation in one affected individual). In addition, as will be discussed in the next section, the excerpt contains a causal argument.

2.3 Scientific Claims and Practice Implications

The focus of our planned annotation efforts is on argumentation for scientific claims and practice implications. In this section we present our analysis of several examples of this type of argumentation, given mostly in the Results section of (Schrauwen et al. 2012). In addition to the instance of Practical Reasoning discussed in 2.2, we analyze Excerpt 2 as making the causal argument shown in Argument 1.

Argument 1.
a. Premise: Several genes in the DFNB93 region are expressed in the human inner ear.
b. Premise (implicit generally accepted assumption): A mutation of a gene that is expressed in a human tissue or system may lead to an abnormality in that tissue or system.
c. Premise: A certain individual (identified as V:14) has arNSHL.
d. Conclusion: The mutations occurring in DFNB93 of V:14 are strong candidates for the cause of V:14's arNSHL.

Argument 1 can be represented more abstractly, for purposes of annotation of similar arguments in the corpus, by the following argumentation scheme. In addition to specifying the premises and conclusion, we have added a critical question. Critical questions associated with an argumentation scheme provide a way to challenge arguments instantiating the scheme (Walton et al. 2008). The use of critical questions in our annotation efforts is discussed in section 3.

Effect to Some Cause in Candidate Set
Premise: There is a causal pathway from G-type events to P-type events.
Premise: An individual has experienced P (a P-type event).
Conclusion: Some G-type event experienced by that individual may be the cause of P.
Critical Question: *What if the set of candidates G does not include the actual cause of the event?*

Excerpt 3 contains the argument described in Argument 2.

Excerpt 3:
"After the identified homozygous variants were filtered through the 1000 Genomes Project November 2010 release and dbSNP131, 47 previously unreported variants remained…" (p. 639)

Argument 2.
a. Premise (same as 1d): The mutations occurring in DFNB93 of V:14 are strong candidates for the cause of V:14's arNSHL.
b. Premise (implicit generally accepted assumption): If a variant is a frequent polymorphism then it is not likely to be the cause of a deleterious condition.
c. Premise: All but 47 of the homozygous variants in DFNB93 of V:14 are frequent polymorphisms.
d. Conclusion: One of the remaining 47 homozygous variants may be the genetic cause of V:14's condition.

Excerpt 4 contains the argument described in Argument 3.

Excerpt 4:
"… 47 previously unreported variants remained and included two exonic mutations, one splicing mutation, six nontranslated mutations, 16 intergenic (downstream or upstream) mutations, and 22 intronic mutations. The two exonic variants included one nonsynonymous variant … in *PFIA1* and synonymous variant … in *GAL3ST3* … The splice-site variant, c.637+1G>T … was located at … of *CABP2* … The variants in *PPFIA1* and *CABP2* were subsequently validated by Sanger DNA sequencing, which only confirmed the splicing variant in *CABP2*. (p. 639).

Argument 3.
a. Premise (same as 2d): One of the remaining 47 homozygous variants may be the genetic cause of V:14's condition.
b. Premise (implicit generally accepted assumption): Only exonic or splice-site variants confirmed by Sanger DNA sequencing could be the cause of a genetic condition.
c. Premise: Of the remaining 47 homozygous variants, only the c.637+1G>T splicing variant in *CABP2* was confirmed.
d. Conclusion: The c.637+1G>T variant in *CABP2* may be the genetic cause of V:14's condition.

Arguments 2 and 3 can be described as instances of the following argumentation scheme.

Elimination of Candidates
Premise: There exists a set of candidates C, one of which may be the cause of event E.
Premise: One or more members of C can be eliminated as candidates.
Conclusion: One of the remaining members of C may be the cause of E.

Excerpt 5 contains two arguments, described in Arguments 4 and 5.

Excerpt 5:
"Next, we checked the inheritance of the *CABP2* variant in the entire Sh10 family … and screened an additional 100 random Iranian controls to ensure that the variant is not a frequent polymorphism. The mutation was not detected in any of the controls, and inheritance was consistent with hearing loss in the family." (p. 639).

Argument 4.
a. Premise: The c.637+1G>T variant in *CABP2* segregates with arNSHL in Sh10 (V:14's pedigree).
b. Premise (implicit generally accepted principle): A variant may be the cause of an autosomal recessive condition if it segregates with the condition in a pedigree, i.e., occurrence of the condition and the variant are consistent with an autosomal recessive inheritance pattern.
c. Conclusion (implicit): The c.637+1G>T variant in *CABP2* may be the cause of arNSHL in Sh10.

Although Argument 4 is in some respects similar to Mills' Joint Method of Agreement and Difference (described in Jenicek and Hitchcock, 2004), its premise (4b) provides a causal explanation that is not part of that type of argument. Argument 4 can be described more precisely as an instance of the following argumentation scheme.

Causal Agreement and Difference
Premise: There exists a set of individuals I-present that have a feature F and property P.
Premise: There exists a set of individuals I-absent that do not have feature F and property P.
Premise: There is a plausible causal link from F to P that could account for the presence of P in I-present.
Conclusion: F may be the cause of P in I-present.
Critical Question: *Is there some other feature G in I-present that could account for P, or is there some other factor G in I-absent that could account for the absence of P?*

Argument 5 from Excerpt 4 is as follows.

Argument 5.
a. Premise: The c.637+1G>T variant in *CABP2* is present in the arNSHL affected members of Sh10.
b. Premise: The variant does not occur in a control group.
c. Conclusion (implicit): The c.637+1G>T variant in *CABP2* may be the cause of arNSHL in Sh10.

Argument 5 can be described as an instance of the following argumentation scheme, based upon Mills' joint method of agreement and difference. Note that its first critical question is shared with Causal Agreement and Difference, but its second critical question is not needed for that argumentation scheme, one of whose premises is that there is a causal mechanism that may account for the differences between I-present and I-absent.

Joint Method of Agreement and Difference
Premise: A set of individuals I-present have a feature F and property P.
Premise: A set of individuals I-absent (distinct from I-present) do not have F and P.
Conclusion: F may be the cause of P in I-present.
Critical questions:

- *Is there some other feature G in I-present that could account for P, or is there some other factor G in I-absent that could account for the absence of P?*
- *Is there a plausible causal mechanism that explains how F leads to P?*

Excerpt 6 contains a causal argument for how the c.637+1G>T variant of *CABP2* could lead to hearing loss, as shown in Argument 6.

Excerpt 6:
"… we evaluated the effect of the c.637+1G>T mutation on splicing … Analysis … revealed … indicating that the mutation of c.637+1G>T leads to a complete skipping of exon 6 … Skipping of exon 6 is expected to lead to a shifted reading frame and a premature truncation of the protein" (p. 639-0).

Argument 6.
a. Premise: The c.637+1G>T mutation of *CABP2* may have a deleterious effect on CaBP2 protein during synthesis by *CABP2*.
b. Premise (implicit): CaBP2 protein plays a role in the auditory system.
c. Premise (implicit generally accepted principle): Damage to a protein can lead to disease of the tissue or biological system in which that protein plays a role.
d. Conclusion (implicit): A c.637+1G>T mutation of *CABP2* may result in a disease of the auditory system.

Argument 6 can be described by the following argumentation scheme.

Cause to Effect
Premise: There is a partially known causal pathway from events of type G to events of type P.
Conclusion: The occurrence of a G-type event may result in a P-type event.

Excerpt 7 contains Argument 7, which is similar to Argument 4 and can likewise be described as an instance of Causal Agreement and Difference.

Excerpt 7:
"We identified two families (Sh11 and He) with affected individuals who were homozygous in this region … Affected family members presented with an audiogram similar to the affected individuals in the Sh10 family… Sanger sequencing … revealed the same c.637+1G>T mutation in these families." (p. 640)

Argument 7.
a. Premise: Affected members of two families, Sh11 and He, have audiograms similar to those of affected family members of Sh10 and the c.637+1G>T variant in *CABP2* segregates with hearing loss in those two families.
b. Premise (implicit generally accepted principle): A variant may be the cause of an autosomal recessive condition if it segregates with the condition in a pedigree.
c. Conclusion (implicit): The c.637+1G>T variant in *CABP2* may be the cause of arNSHL in Sh11 and He.

Perhaps because they expect it to be obvious to the intended audience, the authors do not explicitly state Argument 8.

Argument 8.
a. Premise (generalizing 4c, 5c, 7c): The c.637+1G>T variant in *CABP2* may be the cause of arNSHL in several pedigrees.
b. Premise (implicit generally accepted assumption): A homozygous mutation known to have a certain effect in some families will have a similar effect in anyone who inherits it.
c. Conclusion (implicit): Anyone having homozygous c.637+1G>T variants of *CABP2* may be affected by arNSHL.

Such an argument could be described by the following argumentation scheme.

Induction/Generalization
Premise: P is true of some members S of a class C.
Conclusion: P is true for all members of C.
Critical question: *What if the individuals in S are exceptional with respect to P?*

The conclusion of Argument 8 is needed as a premise of Argument 9 for the practice implications of the article given in Excerpt 8 (which, unlike the other excerpts in this paper, comes from the article's Discussion section).

Excerpt 8:
"In conclusion, we identified mutations in *CABP2* in individuals with moderate-to-severe hearing loss. Mutations in *CABP2* cause an audiometric phenotype that is seen in most families segregating arNSHL. Our results suggest the importance of screening for mutations in *CABP2*, as well as in *TECTA*, in families with this milder audiometric phenotype." (p. 644)

Argument 9.
a. Premise (implicit): The reader's goal is to prevent or mitigate the occurrence of arNSHL.
b. Premise (implicit, same as 8c): Someone having homozygous c.637+1G>T variants of *CABP2* may be affected by arNSHL.
c. Premise (implicit): Screening may determine if someone has homozygous c.637+1G>T variants.
d. Premise: (implicit) Knowing if someone has homozygous c.637+1G>T variants is necessary to prevent or mitigate the occurrence of arNSHL.
e. Conclusion: It is desirable to screen for c.637+1G>T variants in *CABP2*.

Argument 9 can be described as a form of Practical Reasoning.

Practical Reasoning
Premise: Agent's goal is to prevent or mitigate the occurrence of D.
Premise: The occurrence of G may result in D.
Premise: Doing Act may result in Agent's knowing if G.
Premise: Knowing if G is necessary to prevent or mitigate D.
Conclusion: It is desirable for Agent to do Act.

3 Inter-Argument Relationships

The previous section illustrates a chained relationship in Arguments 1-3, i.e., the conclusion of Argument i is a premise of Argument i+1. Arguments 4 and 5 share the same conclusion: *The c.637+1G>T variant in CABP2 may be the cause of arNSHL in Sh10*. The conclusions of Arguments 4, 5, and 7 (*The c.637+1G>T variant in CABP2 may be the cause of arNSHL in Sh11 and He*) in combination support the premise of Argument 8, whose conclusion is: *Anyone having homozygous c.637+1G>T variants of CABP2 may be affected by arNSHL*. The conclusion of Argument 8 is further supported by the conclusion of Argument 6: *A c.637+1G>T mutation of CABP2 may result in a disease of the auditory system.*

To provide an explanation for why the authors chose to provide various arguments, rather than merely observing their presence in the text, we must consider how the authors expect their arguments to be challenged or evaluated by the intended audience. Note that the chain of Arguments 1-3 could be challenged by posing the instantiated critical question of Argument 1: *What if the cause of V:14's genetic condition was not in the set of candidates that were tested?* Rather than directly responding to that critical question, the authors continue with Argument 4 whose claim is that the c.637+1G>T variant is the cause of arNSHL in V:14's family, Sh10. In other words, Argument 4 makes a broader claim, a claim that subsumes the claim of Argument 3.

Argument 4 can itself be challenged by posing its critical question: *Is there some other feature G in I-present that could account for P…?* Then one could explain why the authors include Argument 5, in which the Sh10 family is compared to a control group.

Argument 8 can be challenged by posing its critical question: *What if the individuals in S are exceptional with respect to P?* The biochemical argument 6 that a c.637+1G>T mutation of

CABP2 may result in a disease of the auditory system provides a response to that challenge.

Dialogue games have been used to model argumentation between intelligent agents (McBurney and Parsons, 2009) and in human-human dialogue (Budzynska and Reed, 2012). A dialogue game could be used to represent this aspect of discourse structure in scientific articles. (See Figure 1.) We shall refer to this new game as SDG (Science Dialogue Game). As in the ASD game (Walton et al., 2008), SDG incorporates argumentation schemes and critical questions. The locutions of SDG are Argue (Author supports a claim with reasons Ri), Challenge (Reader requests an argument for a reason Ri given in the author's argument), Pose (Reader requests an answer to address an instantiated critical question of the argumentation scheme of the author's argument), and Reject (Author rejects a hypothesis given elsewhere in the text). Reflecting a writer's reliance on discourse context and expected background knowledge and inferential capabilities of the reader, the reasons of an argument may be implicit in SDG.

The Dialogue Rules of SDG reflect weaker ordering constraints in text than in dialogue and the fact that the reader is imaginary: The permissible replies of the reader to Argue are: implicit Challenge, implicit Pose, or silence. The permissible reply of the author to Challenge or Pose is Argue.

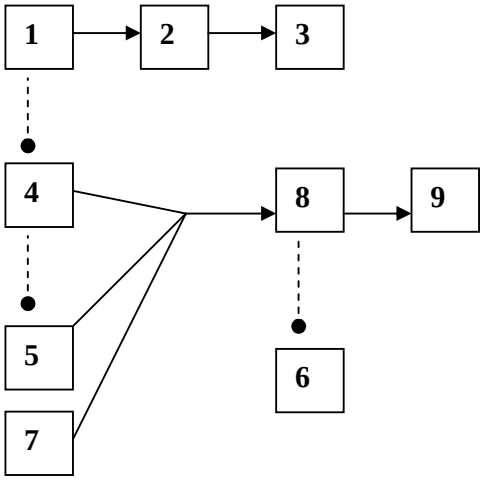

Figure 1. SDG structure. Arrows show conclusion-to-premise support between arguments. Lines ending in circles show responses to critical questions. Conclusions of arguments 4, 5, and 7 are aggregated into premise of argument 8.

4 Discussion

This paper described our analysis of some argumentation schemes and inter-argument relationships in a research article as part of our initial effort to create an annotation scheme. We are continuing to analyze representative articles as preparation for developing and evaluating the annotation scheme. Our longer term goal is to create a freely available corpus of open-access, full-text scientific articles from the biomedical genetics research literature, annotated to support research on argumentation.

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References

Budzynska, K. and Reed, C. 2012. The Structure of Adhominem Dialogues. In Verheij, B., Szeider, S., and Woltran, S. (eds.), Computational Models of Argument: Proc. of COMMA 2012. Amsterdam, IOS Press, 454-461.

Green, N. L. 2014. Towards Creation of a Corpus for Argumentation Mining the Biomedical Genetics Research Literature. In *Proc. of the First Workshop on Argumentation Mining,* ACL 2014, Baltimore, MD.

McBurney, P. and Parsons, S. 2009. Dialogue Games for Agent Argumentation. In I. Rahwan, G.R. Si mari (eds.), Argumentation in Artificial Intelli gence, Springer, Dordrecht, pp. 261-280.

Schrauwen et al. 2012. A Mutation in CABP2, Ex pressed in Cochlear Hair Cells, Causes Autosomal-Recessive Hearing Impairment. *The American Journal of Human Genetics* 91, 636-645, October 5, 2012.

Teufel, S. 2010. *The Structure of Scientific Articles: Applications to Citation Indexing and Summarization.* Stanford, CA, CSLI Publications.

Walton, D., Reed, C., and Macagno, F. 2008. *Argumentation Schemes.* Cambridge University Press.