FRAMEWORKS FOR REASONING AND ASSESSMENT IN MENDELIAN GENETICS

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Step One – Defining Understanding

The first step in developing assessment instruments is to define the nature of the understanding to be assessed. The definition would serve three purposes:

- delineate desired outcomes for instructor and learners;
- guide analysis of learners' ideas; and
- provide a framework for instruction and learning.

When we began our work in 2003, we defined the understanding to be assessed in three ways – being able to:

- *define a concept and identify where it does and does not apply*;
- *use and move between standard representations of a scientific concept;* and
- *use a concept to explain multiple phenomena.*

The first definition of understanding grows out of the classical theory of concepts where concepts are characterized by their necessary and sufficient conditions (Bridgeman, 1927; Carnap, 1955; Jackendoff, 1989; Landau, 2000). The second definition, the prototype theory of concepts, is based on typicality (Smyth et al., 1994; Osherson and Smith, 1981; Lakoff, 1987). Prototypical representations in science include standard diagrams, models, analogies, or mathematical equations. For example, $6CO_2 + 6H_2O \rightarrow C_6H_{12}O_6 + 6O_2$ is a standard way of representing photosynthesis. The final definition of understanding focuses on utility (e.g. Lawson, 2000). What can the learner explain? This grows out of the knowledge-based model of concepts which likens concepts to scientific hypotheses subject to empirical verification or falsification (Wisniewski & Medin, 1994; Murphy & Medin, 1999; Johnson & Keil, 2000; Murphy, 2000).

Our early work showed that our initial definition of understanding served well the first purpose of defining desired outcomes. It was adequate when we used it to guide analysis of learners' ideas. We wrote questions that asked students to work with standard representations or apply a concept to a novel example. (Questions that asked students to define a concept were already readily available.) This led to a list of concepts and associated areas of misunderstanding. For example, for the idea of cellular respiration, students could define the concept and work with many of the standard representations, but very few could use the idea to explain mass loss in organisms. However for Mendelian genetics, specifically meiosis, students had difficulty producing or working with the standard "butterfly" representation of a duplicated chromosome. However the problem was that this diagnosis was not particularly useful for framing instruction. Each problem identified appeared to need its own instructional intervention.

A closer look at students' writing revealed a pattern that crossed topics: many responses violated simple scientific principles. Students' explanations had matter disappearing or appearing out of nowhere or changing in ways that violated basic rules of chemistry.
Energy came and went without appropriate explanation. When looking at genetics-related responses, information in the form of nucleotide sequences was replicated and changed without regard to very basic rules. This led us to be more specific about the type of reasoning we wanted students to use when explaining phenomena or using standard representations. We use the term ***principled reasoning*** to describe a type of understanding where students:

- *provide explanations that are commensurate with a few basic organizing principles as well as knowledge of biological events.*

In particular, we have found that tracing matter, energy, and/or information are useful organizing principles for cellular biology and genetics. By tracing matter we mean identifying the matter that is changing and the chemical identity of the molecules involved, describing the nature of the change, and conserving mass or accounting for mass changes. Tracing energy is a related principle that includes identifying the energy that is transformed and/o transferred and describing the nature of the change while conserving energy. Tracing information is a somewhat less familiar principle which we define as including identifying the nature of the information (nucleic acid or amino acid sequences, gradients, protein conformations, etc.) and how one type of information is translated into a specific signal. For example, the concentration of ATP is an indicator of the energy needs of a cell. High levels of ATP inhibit the allosteric enzyme phosphofructokinase, thus the information in the ATP concentration is translated or transferred to a conformational and activity change in the enzyme.

A Framework for Reasoning

The key ideas for principled reasoning about genetics are outlined in Figure 1. In this framework, the organizing principles of tracing matter and information are columns. Tracing energy is not an important organizing principle for this topic. The rows are levels or system scales. We see biology as a nested set of systems of decreasing scale. The framework shown deals with the subcellular through the organismal levels thus encompassing the content of an entire basic genetics course. An ecosystem level can be added to address population genetics and evolution. Each level is subdivided into the relevant processes. The last column echoes back to the level, identifying more specifically where the processes occur.

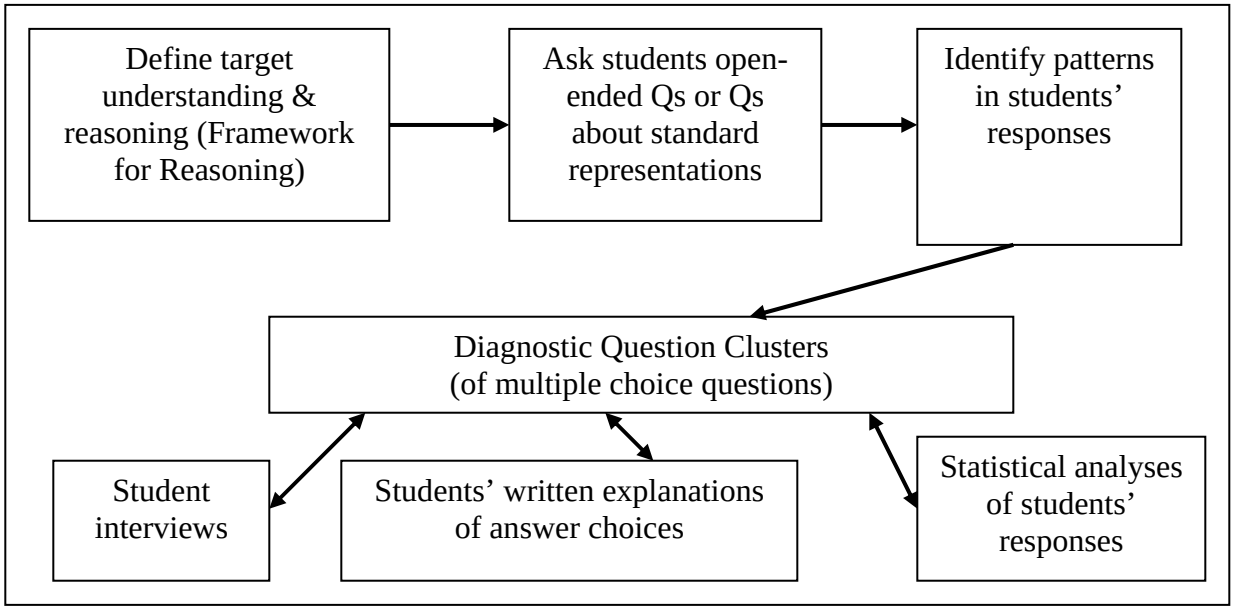
Figure 1. A framework for reasoning about genetics

	Tracing Matter	Tracing Information	Context / Location
ORGANISMAL LEVEL			
Phenotype	Interaction and regulation of **gene products,** beginning with fertilization, throughout the lifetime of an organism – observable or measurable traits.	Genetic information is variably expressed in time and space to "build" a functioning organism	All cells in all living organisms express particular traits
Gameto-genesis	Production of gametes by specific tissues/organs. Number of chromosomes is halved (see *meiosis*).	Genetic information: halved in particular cells.	Occurs in sex tissues/organs of variable complexity from plants, roundworms, flies, etc.
CELLULAR LEVEL			
Mitosis	One copy of each duplicated chromosome/DNA double helix distributed to daughter cells (see *replication*).	Duplicated genotype is distributed equally to daughter cells.	In the nucleus of eukaryotes; in the cytoplasm of prokaryotes.
Meiosis	Each chromosome pair* is halved independently. *in the case of ploidity, there will be more than two.	Genetic info: halved Alleles for each trait are separated following replication and pairing of homologous chromosomes, assuring one member of each pair in daughter cells	Occurs in the nucleus of eukaryotes.
Differential Gene Expression	Set of proteins/polypeptides translated in a cell.	Which ssDNA are templates for transcription is controlled by gene products, DNA modification, etc.	Eukaryotes - nucleus, prokaryotes - cytoplasm. Cytoplasmic interaction of gene products determines cell biochem.
Genotype	Set of nucleotide sequences that comprise the genes/alleles of each cell in an organism. Each allele/gene corresponds to a gene product.	Expression and interaction of allele products, polypeptides, determines trait expression (homo-, heterozygous, etc.)	In the nucleus of eukaryotes, in the cytoplasm in prokaryotes; expressed in cell primarily via translation
SUB-CELLULAR LEVEL			
Translation	Formation of a polypeptide chain, the gene product. Consists of a series of amino acids	Sets of three ribosomal nucleotides = anti-codons for amino acid sequence making up polypeptide chain	Occurs in the cytoplasm.
Transcription	Synthesis of mRNA from nucleotide subunits	ssDNA = information for mRNA synthesis (=allele/gene)	Occurs in the nucleus of eukaryotes, in the cytoplasm in prokaryotes.
Replication	Faithful doubling of DNA (or RNA) from pool of nucleotides in preparation for cell division.	ssDNA is template for formation of new double helix. Changes in nucleotide sequences of pre-gametic cells = mutation.	Occurs in the nucleus of eukaryotes, in the cytoplasm in prokaryotes.

Developing Questions that Assess the Defined Understanding

We use the frameworks for reasoning as ways of focusing and organizing the questions we develop. We aim for "diagnostic" question clusters, since our goal is to identify problems with students' principled reasoning. Figure 2 shows the general method we use to develop diagnostic question clusters. Once we defined the type of understanding and reasoning that we want to assess, we ask students open-ended application questions that require the use of the designated part of the framework and we look for patterns in their responses to these questions. Based on the patterns, we develop multiple choice items where the foils are intended to represent common problematic ways of thinking. We test the robustness of our explanation of why students choose particular foils by interviewing students, having them write about their answer choices, and by doing statistical analyses. Our goal is to develop clusters of multiple choice questions or at least easily gradable open-ended questions, because we deal with large enrollment courses.

Figure 2. Method for developing diagnostic question clusters



Current Work on Question Clusters for Mendelian Genetics and Meiosis

We have developed a question cluster for cellular respiration (Wilson et al, 2006; Wilson et al, in preparation) and are finishing a cluster on photosynthesis. Here we would like to showcase our current work on a cluster for Mendelian genetics, specifically the role of meiosis in explaining Mendel's law of independent segregation. In contrast to photosynthesis and respiration, we find that several of the standard representations are problematic for students. This is especially true when we ask students to keep track of both matter and information. Figure 3 shows an open ended question about meiosis and Figure 4 the patterns that we found among the students who failed to appropriately trace matter and/or information.

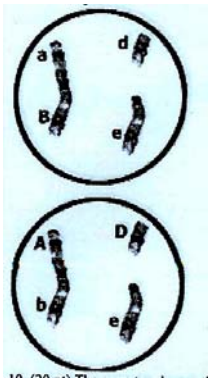
Figure 3. Open-ended question about meiosis

The gamete shown are TWO of the gametes produced when a single parent cell underwent a meiotic division.

A. Since meiosis of a single parent produces 4 gametes, draw a picture of the other Two gametes labeling the appropriate alleles.

B. Draw the parent cell in G1 phase prior to meiosis labeling homologous chromosome pairs and alleles as either dominant or recessive.

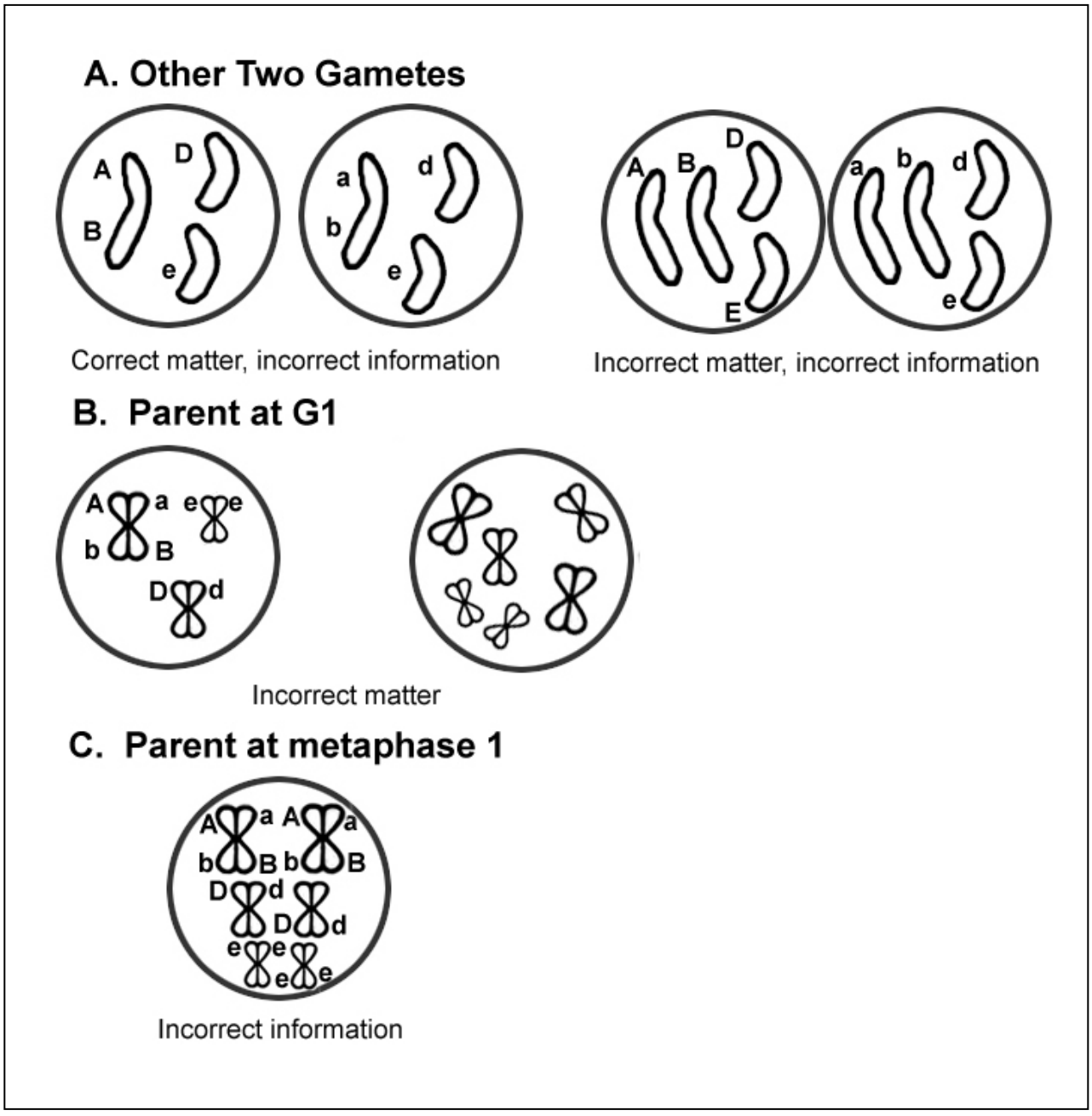
C. Draw the parent cell in metaphase I.



Students in an introductory biology class were given this question on an exam. The variation in their responses was huge. Without a framework it would be difficult to see patterns that could lead to the meaningful development of distractors. However our framework for reasoning suggests that categories such as correct matter (chromosome structure) – incorrect information (labeled alleles) might be useful. Figure 4 shows the most common categories of inaccurate responses to each part of the question. Pictures without designated alleles indicate that no one arrangement of alleles dominated. For part A, 51% of the 108 students drew the correct gametes, while 23% represented the matter correctly but had incorrect information/alleles, and the remaining 26% did not represent the matter correctly. For parts B and C, the examples given represent the most common responses, but not all of the responses.

We are currently drafting multiple choice versions of this question. The distractors for part A will include the responses shown here plus the other variations of incorrect alleles that resemble the first picture in Figure 4. The distractors for part B will include the first picture shown above as well as the many allelic variations of the second picture. Because the vast majority of students who answered part C incorrectly had the chromosome structure correct (paired dyads), all of the distractors will have this format with different variations of alleles. This cluster of questions will also include other multiple choice questions about gametogenesis and meiosis.

Figure 4. The most common incorrect responses to the question shown in Figure 3



Conclusions

We have been studying students' ideas about genetics for many years, but were originally unable to see patterns in their ideas that were useful for framing instruction. For example, it was clear that students needed more help interpreting the standard representations of chromosomes, particularly dyads. Students also confound double helices, homologs, and duplicated chromosomes. Many students who can use Punnett squares to solve inheritance problems cannot identify the possible gametes produced by the parents. These conceptual barriers that students encounter are related, but a unified instructional intervention was not evident until we looked across concepts and identified principled reasoning as a focus for assessment, a way of categorizing students' responses, and framing instruction.