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Medical Diagnosis and Actual Causation

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ABSTRACT. I suggest that some diagnoses can be seen as causal explanations based on "particulars" – instead of regularities – and on the notion of actual causation. Diagnoses based on case-based reasoning provide a particularly vivid example.

1. Medical Diagnosis and Explanation

Diagnostic problem solving is usually a complex procedure, which involves many steps and a plurality of cognitive and inferential activities. Once formulated, however, a diagnosis can be synthetically described, from a statistical viewpoint, as a *relation* between a set of findings (signs, symptoms, laboratory test results) and a certain pathological condition attributed to the patient. What kind of relation? According to a common opinion among experts in computational models, medical diagnoses express *explanatory* relations: for example, Reggia, Nau, and Wang (1983) state:

> […] diagnostic reasoning involves a sequential hypothesize-and-test processing during which the physician conceptually constructs a model of the patient. The "hypothesis" postulates the presence of one or more diseases that could explain the patient's manifestations.

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The claim that all diagnoses are explanations is not unanimously shared: Federspil (2004) distinguishes between nosographical and physiopathological diagnosis and accepts as ‘explanation” only the latter, which aims at reconstructing mechanisms which have brought about the actual conditions of the patient and have a definite causal character; nosographical diagnosis would have instead only a classificatory character.

It might be answered that in a classical treatment of explanation also nosographical diagnosis might be seen as explanation, only “non causal”. Hempel (1965, pp. 453-7) admits that subsumption under a concept can be explanatory, but only when the concept points to some kind of regularity, as in medical diagnosis. However I think that a characterization of medical diagnosis *à la* Hempel would not be fully satisfactory for two reasons: in some diagnosis the explicatory work is not given by regularities; besides, in such diagnoses, causality has a more important role than the one given by Hempelian models.

2. Regularity-based Explanations

In the inferential and regularist Hempelian view, the explanation of an individual event – for instance that a person has a certain set of signs and symptoms (S_i) – is given when the event can be deduced by a set of laws and particular facts. But in medical diagnosis we have weaker relations than deduction, and diagnoses have generally a probabilistic character. Diagnosis therefore is given when we can sort out the pathology which is the best explanation of the patient exhibiting S_i. In a Bayesian approach, the best explanation is the pathology which appears more probable given S_i, where the probability of each diagnostic hypothesis is computed with the Bayes formula. Such an approach permits to evaluate how the acquisition of new evidence changes the probability of different diagnostic hypotheses, facilitating comparisons among rival hypotheses. Bayesian diagnostic reasoning is made easier by the use of computational models, like Bayesian networks (see Pearl 1988, Pearl 2000, Spirtes, Glymour, and Scheines 1993). A Bayesian network is a directed graph in which nodes represent statistical variables, edges represent conditional probabilities, and no cycles are allowed. Various assumptions, among which Markov assumption, allow uses of Bayesian nets as inference tools.

Bayesian networks may have a probabilistic interpretation, and therefore model merely nosographical diagnoses, or a causal interpretation, and therefore represent possible physio-pathological mechanisms that could have produced S_i. Diagnostic procedures devoted to get the most probable hypothesis use *type* probabilities: the patient is seen as member of a population.

Expert systems that use causal Bayesian networks make appeal to the notion of probabilistic causality (see Long 1989), whose basic idea is that A causes B if A increases the probability of B. In both probabilistic and causal interpretations, the relations of probabilistic independence given by graphic models are relations among event types, and causal relations are relations of type causation. Therefore both probabilistic and causal diagnoses obtained through Bayesian Networks are based on *types* and on statistical *regularities*. This is no surprise, and nobody wants to cast doubts on statistical data and their role in clinical sciences. However, if we admit that some diagnoses aim, at least ideally, at reconstructing the process that produced a set of signs and symptoms in an individual patient, pointing towards a causal explanation of the patient's individual condition, we may see that an explanation based on regularities and on general causal relations cannot be adequate. Probabilistic regularities and relations actually do not always *explain* individual cases.

When a physician examines a patient who exhibits a symptom, she does not wants to discover what causes that symptom in general, or in humans: she wants to discover what caused the presence of the symptom in her patient. Causal diagnoses based on general knowledge and on regularities must confront with the fact that sometimes the most probable cause of a conditions *in humans* is not the *actual* cause of that condition in a *given patient*. The notion of *actual causation*, recently much debated among philosophers, could reveal more useful. But this notion is not based on regularities.

3. Clinical Explanation and the Problem of Individuals

The idea that diagnosis can be modeled as an explanation based on the study of individuals has been often considered in the philosophy of medicine. Gorovitz and MacIntyre (1976) stress the fact that medical science is characterized as *science of particulars*:

> every particular continues to exist and has the characteristics that it has only in virtue of the operation of some set of physical and chemical mechanism. Some particulars – ice cubes and molecules are notable examples – are such that nearly everything that we might want to know about them can be explained simply by citing the relevant mechanisms.

Other particulars, however, like the higher primates, "cannot be understood solely as the sum-total of the physical and chemical mechanisms that operate on them. What effects such mechanisms do have is affected by the particular history of that specific particular with all its contingent circumstances, contingent that is, and even accidental, relative to the operation of the mechanism." Gorovitz and MacIntyre want to stress that knowledge of individuals as given by medical science keeps always a degree of uncertainty, which does not come from an imperfect knowledge of regularities or initial conditions or both; on the contrary, the uncertainty derives from a necessary "ignorance of the contingencies of the environmental context".

Schaffner (1986) has discussed the relation between the role of generalizations and the study of individuals in biomedical sciences. He claims that "the nature of biomedical theorizing requires an increased emphasis on what might be called *prototype organisms* and on *analogical extension of biological knowledge*" (1986, p. 68). Schaffner's main point is that many

> biomedical theories lack generalization of broad scope", focusing instead on "an appropriate organism- often referred to as an 'animal model'" and "articulates a mechanism […] that explain some feature(s) of that organism. […] The organism (or component part of the organism) is construed to be a prototype, and to have similarities with other organisms […] that license the extension of the 'mechanism' to a broader class of biological entities. Such extension is typically analogical […] (Schaffner 1986, p.70).

Schaffner gives some space also to clinical sciences, considered as the area where the "exemplar thinking" emerges with particular clearness.

4. Case-based Diagnoses as Explanation based on Individuals

An application of analogical reasoning to diagnosis which is gaining increasing attention in health sciences is given by case-based reasoning (see Ankeny 2006 and Bichindaritz 2008). Case-based reasoning is usually defined as a methodology of problem-solving based on the analogy to already solved similar problems. In clinical applications, case-based reasoning (CBR) starts by construction of an "index case", i.e. a description, in more or less detail, of a particular case, often a report on an individual patient (Ankeny 2006); when a new diagnostic problem is presented, the case base is searched in order to find the best matching case(s) among the retrieved ones; the assessment of the similarity relations between the two cases can induce a modification of the index case (possibly by generating a more abstract index case) and/or the adoption of the new case as index case. In computer science, the standard description of CBR is the following:

> It basically consists in *retrieving* past cases that are similar to the current one and in *reusing* (by, if necessary, *revising)* past successful solutions; the current solved case can then be *retained* and put in the system knowledge base, called the *case base* or the *case library*." (Montani 2008)

Even from these minimal sketch it should be clear that CBR is based on individual cases (and analogies); it is true that often similar cases are used in order to obtain a generalization, an abstract prototype; but generalization is not the starting point of the problem solving procedure; rather, when applied to diagnostic problems, CBR appears as a model construction, where some individuals, or their clinical history – the retrieved cases - become the model for new cases.

5. Case based Diagnoses and Actual Causation

Answers to the question whether one particular event caused, or prevented, another, are judgments of *actual* causation. The peculiarity of actual causation emerges in situations of causal *pre-emption,* i.e. situations in which two configurations of factors are present and both are nomically sufficient to bring about an effect, but just one of them actually causes it. For example, suppose that a patient has a leg edema (LE) and that it has been caused by a kidney problem (K), but our patient has also a cardiac problem (H) that would have nevertheless caused a leg edema.

As it is well known, causal pre-emption has raised problems both to regularity and counterfactual accounts of causation; however, here we consider just the counterfactual account, as it is the most appropriate to singular causation. In the case of our patient, there is a "causal situation" in which both K and H are causes of LE, but we have the intuitions that K is the actual cause of it. LE does not depend counterfactually on K, nor is K a proper difference-maker w.r.t the patient's situation, as LE would have been present anyway[1]. In

[1] Obviously the cure should try to fix both K and H, but in many context the correct identification of the causes of the patient's symptoms is important.

more promising recent works, actual causation is presented as counterfactual dependence which holds not in the actual situation, but “in a certain kind of ‘normalized’ version” of it (Hitchcock and Knobe 2009). In our example, K causes LE in our patient if both K and LE are present, and if in a virtual, “more normal”, situation in which the patient has no cardiac problem, his LE would counterfactually depend on K. Therefore the notion of actual causation seems strictly related to some standard or notion of “normality”. But what is normality? Hitchcock and Knobe (2009, p. 598) stress that the notion of normality is context- dependent, and that there are different systems of norms: beside statistical norms, we find moral norms, and also “norms of proper functioning that apply to artifacts and biological organisms (and their components)”.

What has all this to do with diagnoses and CBR? Our suggestion is that those diagnoses which are, to some extent, causal, aim to detect the actual causes of the patient’s condition. These causes can be characterized as difference-makers wrt. the ‘normal’ situation of the patient. But in this case, normality does not coincide necessarily with statistical normality (otherwise, improbable causes would be condemned to eternal ‘inactivity’). Rather, each individual is better characterized as the ‘proper’ functioning of a model which is similar to the individual in question under relevant aspects.

Diagnosis via CBR can be seen as explanation based on difference from a norm of “proper functioning”: whenever an index case is inserted in a case base, it is inserted because it presents some abnormal features with respect to the “normal” abstract organism, i.e. a properly functioning organism. However, when compared with a new case, it is the index case – as a prototype of abnormal functioning – that dictates the ‘norm’.

6. Conclusions

Case-based diagnosis seems to express a form of causal explanation linked to actual causation. This kind of diagnosis is not necessarily an alternative to other form of diagnoses, like those based on Bayesian probabilities; rather it is conceivable to apply ordinary Bayesian reasoning in standard problems and CBR in situation which appears to be nonstandard, as rare pathologies (see Montani 2008). In any case, CBR diagnosis offers an example of diagnosis that is compatible with the most recent idea of causal explanation based on actual causality.

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