

## Medicine of the Person

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# Real, virtual and potential illnesses: an epistemology of care

## Diagnosis versus care

Diagnosis and treatment are the two main fields of medicine. Historically, the request for treatment preceded and generated the diagnosis, and getting the diagnosis right contributed to making the care better.

Nowadays, there is an ever greater dissociation between these two areas:

- The improved accuracy of diagnosis is having less and less of an effect on the quality of care
- Patients place at least as much importance on obtaining the diagnosis as they do on getting treatment.
- Doctors have a capacity for diagnosis which far outstrips their capacity for treatment.
- The choice of treatment is subject to multiple media, medical, commercial and political pressures.
- Making the diagnosis has been taken out of the hands of clinicians and is getting further and further from the real health of individuals.

The epistemology of care encompasses the study of its different modalities, of its objectives, of its subjective results (both for patients and doctors), of its criteria for evaluation (biases, thresholds), of its objective results in terms of quantity and quality of life, and of its various anthropological aspects (empathy, cooperation, family), sociological aspects (commercial, social groups, rituals, hierarchies, biomedical capabilities), technological aspects (prostheses, resuscitation), psychological (placebo effect) and cognitive aspects (information, understanding, adherence).

In order not to get lost in such an enormous subject, I would now like to propose a classification into three big groups of diseases, not by aetiology, type or gravity, but solely by the degree of dissociation between the accuracy of the diagnosis and the quality of care given.

## Illness

An individual's health invokes a subjectivity which cannot be reduced simply to science. There are as many definitions of health as there are subjective experiences of health, that is to say individual people. Faced with this definite impossibility of defining health, science addresses exclusively those 'objects of study' which are the diseases. Whether an individual considers his health to be excellent, poor, precarious, variable, average, fragile, mediocre, or faltering, gives absolutely no idea of the nature or number of 'object-illnesses' they suffer from. None of the debates about the difficult link between medical science and the humanities or between doctors and patients can be understood or analysed without this basic premise: health is 'subjective', illness is 'objective'.

Just like 'so-called planets' whose definition has varied with the state of our knowledge, 'so-called diseases' have a reality which translates the state of knowledge in a given age and allow the dissemination of knowledge.

So-called illnesses, defined nowadays by biomedical science have little in common with the so-called illnesses of yesteryear, arising as a result of a request for treatment. We will now examine three big classes of so-called illness, two of which are very recent.

### Real illnesses

'Clinical episodes' consist of almost infinitely variable symptoms (pain, weakness, breathlessness, cough, reddening, anxiety, bleeding, fracture, etc.). 'Clinical events' are the symptoms and incidents which the doctor writes in a clinical history with the aim of establishing a diagnosis.

The four criteria which define a real illness are the following:

- The initial clinical episode is lived by the patient (or his companions)
- The patient (or one of his relatives) goes to the doctor to tell them about it
- The doctor considers it to be a clinical event
- Biomedical science makes a diagnosis linked to the clinical event

Real illnesses are the 'classics' of medicine (colitis, migraine, appendicitis, stroke, tuberculosis, fractured tibia, schizophrenia, temporal epilepsy, rheumatoid arthritis, urinary calculus, whooping cough, Parkinson's disease, etc.): they encompass almost the whole ICD (international classification of diseases).

The link between the clinical event and the biomedical diagnosis is direct, which isn't always the case with the two other classes of illness which we will consider.

These real illnesses are not unique to homo sapiens. All types of animals have real illnesses (parasitic, traumatic, congenital or genetic), even if they don't go to the doctor to get a diagnosis. One can arrive at an estimate for each species. Other than catastrophe, famine or epidemic, it is rare for more than 5-10% individuals to suffer from real illnesses. Real illnesses are inherent in the laws of nature and of evolution.

In man (and domestic animals), psychiatric pathology and pathologies linked to ageing are also real illnesses, since there is concordance between the experience lived by the patient and the biomedical diagnosis.

The influence of medicine on real illnesses is evaluated using QALY's (quality adjusted life years). We should remind ourselves here that premature death is defined as death before the age of 65. Medicine has contributed to the spectacular reduction in premature deaths but it has had a very modest or mediocre effect on non-premature deaths.

Historically, real illnesses are the *raison d'être* of medicine with its excellence in diagnosis, and its growing successes in treatment (vaccines, surgery, vitamins, prostheses, dialysis, insulin, antibiotics, etc.)

### **Interlude on the absence of diagnosis**

Before embarking on the group of virtual illnesses, we must avoid any risk of confusion with a subject which is much more extensive (and outside our remit). I'm talking about the absence of diagnosis, expectant diagnoses, diagnoses of convenience or rubbish diagnoses. All of these negative terms to describe those situations where biomedical science fails to find an explanation for the clinical episode or the suffering of patients. In short, those times when medicine fails both diagnostically and therapeutically. This goes beyond the classical context of psychosomatic illnesses and hypochondriasis, since, in spite of all the progress in investigations, there are still acute or chronic pain, chronic fatigue, sensory deficits, temporary weaknesses for which medicine has absolutely no diagnosis to propose to the patient.

In certain cases, such as period pains caused by endometriosis or the diurnal fatigue of obstructive sleep apnoea, a biomedical diagnosis was, for a long time, lacking. For the most part, medicine has fallen into the habit of abandoning the psychosomatic and filling the void with all those infamous provisional or contested diagnoses, sometimes referred to as rubbish diagnoses: in the past spasmophilia (a condition in which only moderate mechanical or electrical stimulation produces spasms, convulsions or tetany) or chronic brucellosis and nowadays fibromyalgia, chronic Lyme disease or gluten intolerance.

All those clinical cases which Molière would have classified as imaginary illnesses are neither real illnesses, for they don't have a biomedical diagnosis nor are they virtual illnesses, for they have clinical symptoms. We are therefore going to speak now of virtual illnesses whose main characteristic is that they don't give rise to clinical episodes.

### **Virtual illnesses**

A virtual illness is not lived by the patient but it has a concrete reality for biomedicine. It is therefore the opposite of an imaginary illness. There is no clinical episode which prompts the patient to consult a doctor.

These virtual illnesses correspond approximately to 'risk factors' and to states that are designated preclinical or subclinical. All cancers detected by screening, arterial hypertension, type 2 diabetes, the gene mutations BRCA1 and 2, arterial aneurysms are 'anomalies' where the recorded abnormality has never given rise to any symptoms experienced by those suffering from them.

Virtual illnesses contain within them a great paradox; the diagnosis is assigned to an individual, whereas what it is really about is a population diagnosis. They are the consequence of the supremacy of medicine driven by probability.

At the individual level, the link between a risk factor and an illness is stochastic rather than determinist, for there is a huge variation in cofactors and life events.

An arterial aneurysm increases the risk of haemorrhage, hypertension increases the risk of cardiac failure or a stroke, a BRCA mutation increases the risk of breast cancer, and a colonic polyp increases the risk of cancer of the colon. But the progression of one to the other is determinist at the population level and stochastic at the individual level.

Biomedicine is uncovering more and more virtual illnesses, but remains incapable of predicting how they will develop in individuals. An aneurysm might never burst, a genetic mutation might never translate into a phenotypic change, a localised cancer might remain for ever localised or even disappear. A virtual illness may never shorten the quality adjusted life years for any given individual.

This stochastic property is demonstrated in a particular way at the level of pharmacological treatments for risk factors; most of them are ineffective before the first clinical presentation (primary prevention) and only show themselves effective afterwards (so-called secondary prevention). There are abundant examples of this (statins, oral hypoglycaemics, antithrombotics, anticoagulants, treatments for osteoporosis, etc.); it is only after the actual illness (stroke, heart attack, fracture) that the treatment becomes effective. Thus confirming the relevance of biomedical science to real illnesses and its persistent naïvety when it comes to virtual illnesses.

### **The reality of virtual illnesses**

But oddly, the evolution of medical and social practises has finished by conferring a clinical reality on these virtual illnesses which is completely new. Virtual illnesses generate a pathological experience which is sometimes farcical.

- What is wrong with you, sir?
- My cholesterol. My PSA. My polyps.
- What is wrong with you, madam?
- My hypertension. My sugars. My abnormal smear.
- No, I'm asking you what really troubles you?
- But I've just told you.
- No, but what is really bothering you?
- Nothing. Apart from that, I'm fine!

All my colleagues have had similar conversations and it is a sign that patients have completely assimilated their virtual illness. These virtual illnesses end up acquiring a clinical importance and an effect on the patient's well-being which is as significant, maybe even more so than their chronic bronchitis, their migraine or auto-immune disease.

Biomedical power has managed the feat of transforming population statistics into cognitive and clinical realities for the patients, and the resultant morbidity may sometimes be greater than that of a real illness! A woman who has discovered her breast cancer by feeling it herself is often less anxious than a woman whose cancer has been detected by screening. The first woman has a clinical knowledge which reassures her as long as she feels nothing, the second is terrified at the idea that anything could happen, even if she feels nothing. Headaches are not the symptoms of hypertension, but a hypertensive will be more anxious if he has headaches, since he knows that it could be a symptom of malignant hypertension. These two examples, among many hundreds, show us that the lived morbidity of a virtual illness may be more onerous than the lived morbidity of a real illness. So it is biomedical science which has generated an excess of morbidity.

Is the doctor's role to create morbidity?

One could riposte that this morbidity that is created is different in quality to the morbidity of a real illness. This argument is not allowable if one does not know the life expectancy of the real illness and if one can't define the impact of that artificial morbidity on other parameters. In short, as long as we don't know the natural history of these virtual illnesses, we will not be able to quantify the QALY's lost or gained by their detection and treatment on an individual level.

Treating the individual is the art of the clinician, but we give less and less weight to this. The time available to talk to the clinician is counted in minutes per year whereas that of the market and the media is counted in hours per day!

In contrast to real illnesses, animals are never bothered by virtual illnesses, since these require cognitive assimilation which is unique to the human species.

Let us try to estimate the percentage of individuals affected by virtual illnesses in developed countries. Considering the politics of screening and the medicalisation of society by the progressive lowering of thresholds of risk, it is not an exaggeration to say that as I am writing these sentences, more than 30% of the adult population are carriers of at least one virtual illness and certainly greater than 60% over the age of 65 (defined as the threshold for premature death).

## Second interlude concerning discordance

Having discarded imaginary illnesses and the rubbish-diagnoses from our argument, we must also get rid of those cases, more and more numerous, of discordance between the clinical symptoms and the proposed biomedical diagnosis. As soon as a doctor can't manage to make a link between a clinical symptom and the list of possible diagnoses, he has two diametrically opposite choices. Either a provisional admission of failure, or flight in the face of limitless para-clinical investigations...

Personal pride, fee for service payment, free healthcare, the mirage of technology and the risks of litigation all contribute in a big way nowadays to people choosing the second option. A particular investigation will necessarily result in one of the following three outcomes:

- The discovery of a positive result defining a virtual illness
- A false positive. The risk ( $\alpha$ ) of a false positive is classically 5% for each analysis. The probability of a false positive increases as a function of the number (n) of tests done; <sup>1</sup> For greater than 40 tests, this probability is 90%!
- An artefact, or an 'incidentaloma' (biliary cyst, diabetic renal disease, etc.) without any individual or population-wide determinism.

In the three cases, the conclusion has no relation to the initial request for care. A patient consults a doctor because of itching or a migraine and leaves with a suspicious naevus, an aneurysm or an 'incidentaloma'.

This has been well-summarized by Rosenberg; 'the demands for technological ingenuity and patients' impatient demands echo in practical terms society's expectations of medicine.'<sup>2</sup>

## The nature of death

In order to understand the third class of illness, that is, the potential illnesses, we must approach death as a means of maintaining, adapting and evolving the species, as important as reproduction (sexual or not).

As seen by a biologist, death is a natural phenomenon whose stages are becoming better and better understood. The number of cellular divisions cannot exceed 50-60 (Hayflick's limit), that limit also applying to the stem cells of each tissue or organ. The mutations which are inherent in each division end up accumulating to an extent which exceeds the capacity for DNA repair by the telomeres. The cells are also damaged by extrinsic agents; ultra-violet radiation, natural radiation, chemical products. Oxidative stress consists of the production of free radicals which are the degradation

products produced as a result of the generation of energy by mitochondria from oxygen. Finally, and above all, illnesses associated with ageing, basically neuro-degenerative, cardio-vascular and cancerous, are not selected out by evolution, since they occur after the age when reproduction occurs.

But, as the doctor writes up the death certificate, he is obliged to mention one or more diagnoses for the medical causes both direct and indirect of death (renal, cardiac or respiratory failure secondary to such or such cancerous, vascular or neurological pathology). Seen from the medical point of view, death is always the result of an acute illness or an incident which exacerbates chronic disease. In medicine the concept of 'natural death' does not exist. This can end up appearing ludicrous, but since medicine has as its goal increasing the quantity and quality of life, and since it is judged by its results, it can't manage by itself to set a limit to this quest for a diagnosis. Biomedical science cannot integrate the idea of death as a 'natural process', without risking a severe identity crisis.

Even if each doctor has their own opinion on the subject, it is not from biomedical science that we should demand the acceptance or management of death; that would be quite simply an oxymoron. The difficulty we have putting palliative care into practice and the impossibility of calmly approaching the questions of euthanasia or of assisted suicide are the most concrete proofs of this.

So, it is the general population and leaders in the field who need to take responsibility for the acceptance and the management of death. But if the citizens of developing countries can accept death, omnipresent in their social landscape, citizens of developed countries are no longer capable of doing so, since we have removed it from their field of view. Most of them will die in an institution or in a hospital. Held to ransom by hubris and by communication which is more and more biased, biomedical science finds itself alone in the face of the impossibly tangled skein which it has created.

From where, even more bizarrely, we arrive at the final class: potential illnesses.

## **Potential Illnesses**

We can define potential illnesses as an unlimited extension of virtual illnesses, resulting from a growing understanding of risk factors and physico-pathological scenarios. This understanding is the result itself of the huge progress in the technologies used in investigation (imaging, microscopy, molecular biology, genomics, metagenomics).

Risk factors which we have more or less assembled into virtual illnesses are infinitely expandable, thanks to constant progress in investigative techniques.

If Mr X is going to die at the age of 85 of cancer of the colon or of a stroke, he will certainly carry several markers for these illnesses decades before the first clinical symptoms appear. Progress in investigative techniques, as well as the prime importance of statistical correlations imposed by evidence based medicine on clinical medicine, allows us to affirm that the predictive disease markers for all pathology will one day soon be detectable from birth onwards.

Among the examples of this uncontrollable drift which have been already caricatured, we must mention individual gene analysis proposed by IT giants to describe simple DNA polymorphisms liable to be correlated with an infinity of pathologies. Already almost one hundred DNA or genetic markers for Alzheimer's disease have been the subject of 'official' publications! What is certain is that each of us will have at least one genetic marker for Alzheimer's disease detectable as soon as we are born.

These potential illnesses are all the cardio-vascular, neuro-degenerative, cancer or auto-immune disease which cause all the non-traumatic deaths in our countries, since infectious diseases have been more or less dealt with. For each of us, our potential illness is therefore one of these four causes of death. This isn't a terribly original thing to say. In contrast, what is becoming unique, is that biomedical investigations allow us to elaborate the diagnosis from the moment of birth.

And so, from the youngest of ages, and in a future which is not that distant, 100% of people will be carriers of between one and four potential illnesses, one of which will prove fatal!

### **What is the problem?**

Let us imagine this future where each person will receive, at a very young age, the diagnosis of the illness which will kill them.

Will this news of the diagnosis have the same potential to artificially create a lived morbidity as the diagnoses of virtual illnesses? (Let us remember that a cancer which has been detected by screening, therefore preclinical, can do as much damage psychologically as a cancer which is clinically evident). Or in contrast, will the diagnostic madness engendered by our scientific progress perhaps lead people to take their medical destiny back into their own hands?

I wouldn't know how to reply to that question.

Those doom-sayers will see in this diagnostic folly the creation of an infinite morbidity perpetuated by an all-powerful market on a subservient people.

The transhumanists will see this as evidence of real progress bringing us ever closer to the dream of immortality.

Personally, as I am neither a catastrophiser, nor a transhumanist, I don't see a major problem with this over-medicalisation which will finish up with 100% people being patients. On the one hand, illness, in becoming the norm, will lose its morbidity; on the other hand, humanity has already had to deal with problems much greater than that of over medicalisation.

The main problem lies elsewhere. It concerns new social realities primarily in the economic and political realms.

In the 1950's and 60's, we progressively passed from an economy of demand to one of supply. Why would medicine, which is a human activity have escaped the evolution of human society?

Medicine, also, has moved from an economy of demand to an economy of supply. Care-giving which used to rely mainly on the principle of sharing and solidarity is in the process of becoming a market economy. Even the realm of vaccination, which for a long time has been spared by the market, is currently contaminated. The inequality of care has already started in a big way in countries such as the US and it is arriving at speed on our shores. We should therefore expect a deterioration of health of the most deprived which will end up having an impact on health indicators.

As they turn the ethics and the weaknesses of political orators for their own ends, those who prey on the health market force the politicians, who wish to keep to the principle of solidarity, to increase taxes. In the end this has repercussions on the economy and on unemployment and therefore also on health indicators. The indecent price of new cancer medicines has a negative impact on health measures which are far more profitable in terms of QALY's.

Fee for service diverts doctors towards virtual and potential illnesses, which are less time hungry than real illnesses.

The irrational fear of litigation diverts doctors from real illnesses where their responsibility is more immediately apparent. Rheumatology, radiology, dermatology or endocrinology will be more attractive in the future than paediatrics, infectious disease, anaesthesia or obstetrics.

The biggest problem both now and in the future, of this creation of virtual and potential illnesses is certainly its negative impact on the treatment and prognosis of real illnesses.

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<sup>1</sup>  $[1 - (1-\alpha)^n]$

<sup>2</sup> Charles E. Rosenberg. *The tyranny of diagnosis: specific entities and individual experience. The milbank quarterly* 2002