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The more advanced the diagnostic methods at our disposal, the more difficult it can be to delimit the diagnosis

The quest for the diagnosis

The young patient had undergone a comprehensive assessment. Clinical, biochemical and radiological findings showed that something was wrong, but the condition did not fit into any medical diagnostic category. When discharging the patient for the second time from the hospital, I had lengthy conversations with the patient and next of kin. The treatment was effective, and the patient accepted that we had no diagnosis. Not so, however, the GP. In a letter to the department, he wrote: «In this modern age, it must be possible to provide the patient with a proper diagnosis.»

Diagnosis is important in clinical medicine. It gives an indication of characteristics shared by a number of patients, and thereby of prognosis and the anticipated effect of treatment. Diagnoses make it possible to plan health services, distribute health resources, monitor the health of the population and carry out preventive work. From a global perspective, diagnoses and diagnostic categories enable comparisons of health, morbidity and causes of death, and distribution of health resources. The way in which we choose to categorise diseases has consequences.

Diseases may be categorised in many ways, for example according to what causes them (aetiology), where they cause symptoms (anatomy) or how they occur (pathophysiology). One of the earliest systems of disease classification is Carl von Linné's (1707–1778) *Genera morborum* (1). Linné classified diseases into 11 categories, mainly based on symptoms but also on what were assumed to be their causes. William Farr (1807–1883) later developed a system of classification based on the organ in which the disease manifested itself (2). This system was further developed by Jacques Bertillon (1851–1922) and in 1899 it was recommended that it be used throughout Europe (2) – at a meeting in Kristiania, as it happens. This was the initial impetus for the International Classification of Disease (ICD), which is currently in its tenth version (ICD-10). ICD-10 is now used in more than 100 countries and is published in 43 different languages. A full 70 % of world health resources are now distributed using ICD-10 (3).

There are several lines stretching from Linné to ICD-10. One of them concerns compromises between various principles of classification. Linné classified in part according to symptoms and in part according to causes. This fundamental confusion is still evident in ICD-10, which partly has classifications based on pathophysiology (for example tumours), partly on aetiology (for example infectious diseases) and partly on anatomy (for example diseases of the respiratory organs).

Of course, in an ideal world it would be desirable for all disease classifications to be based on aetiology. The rapid developments over the last few decades in areas such as genetics and imaging diagnostics gave rise to the hope that purely aetiologically based

diagnoses would finally become a reality. Paradoxically, the opposite has happened – it transpires that the more biomarkers for disease we have, the more difficult it is to delimit the diagnostic entities. The same spectrum of genetic risk factors and biochemical markers is often discernible in many diseases, particularly in the field of psychiatry (4). This makes diagnostic delimitation more difficult, not easier, than previously.

It is therefore no surprise that the revision of ICD-10 is a prolonged process. The next version, ICD-11, has been postponed until 2017 (3). The work involves medical professionals, patients, health authorities and other actors throughout the world. Anyone at all can comment on a provisional version available on the internet (5). The work can be followed on Twitter at @whoicd11.

The way in which diseases are categorised has consequences. For example, it is likely that stroke will be moved from diseases of the circulatory system to diseases of the nervous system (5). Stroke is the world's second most frequent cause of death and the third most frequent cause of disability (6). When such a large disease group is moved, there are consequences for morbidity and cause of death statistics. The medical field that «owns» a significant disease will be able to claim that they are entitled to more resources.

Diagnoses vanish and new ones appear, often in line with fluctuating diagnostic traditions. Psychiatry is a good example of this. At other times, new diagnostic methods alter the diagnostic landscape, as is the case when prion diseases and paraneoplastic syndromes receive their own diagnosis blocks in ICD-11 (5).

Data from the UK have shown that the ICD-10 chapter *Symptoms, signs and abnormal clinical and laboratory findings not elsewhere classified* is the most common diagnostic group for consultations in general practice (7). A Scandinavian study showed that general practitioners gave the same patient with unspecific health complaints up to 31 different diagnoses (8), and at many hospital outpatient clinics, medically unexplained symptoms are the most frequent diagnosis (9). Despite detailed diagnostic systems, many of our diagnoses are still uncertain and poorly delimited. There is no reason to believe that this will change significantly in ICD-11. The world we are attempting to categorise is too complex for that.

The more advanced our diagnostic methods become, the more the individual diagnostic categories appear to slip away from us. Thus the reply to my patient's involved GP had to be: No, sometimes it is not possible to provide a proper diagnosis, especially not in this modern age.

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