The utility of psychiatric diagnosis may be overstated, or it may be disparaged, depending on one’s point of view. So, it is appropriate to consider what a psychiatric diagnosis really is. It is a fallacy to think that all the so-called psychiatric disorders are meaningful entities, from a classical medical, that is, an etiopathogenic, point of view. It fact, none of them are. Whatever validity they have is based on no more than natural history, genetic associations, or response to specific treatments.

There has been so much criticism of the DSM-3s that it is almost mendacious to assert that there may be some intelligence behind the thing, too that it is to advance the hegemony of psychiatrists and to fatten the coffers of their professional association. The DSM-3 and its baby sister, the DSM-3r, are composed of nothing more than systematized descriptors that borrow heavily from the research traditions of the past few years. The diagnostic entities contained therein are nothing more than phenomenological constructs, and their purpose is as much to improve communication and understanding among researchers as it is to systematize clinical practice. It says nothing about etiopathogenesis, which is what most people suppose medical diagnosis is really about. It says nothing about pathophysiological mechanisms that may be manifest in different ways among people with different levels of cognitive ability.

The legitimate question, of course, is not how mentally retarded people may be stretched or shrunk into a Procrustean bed of DSM categories, but to what degree, if any, those categories have any useful relevance to understanding and managing certain individuals who also happen to be mentally retarded. Or to what degree it is a useful intellectual exercise, something that advances medical science. And to what degree it is a useful clinical endeavor, something that will advance the understanding and the treatment of some individuals and not simply justify, post hoc, the administration of psychotropic drugs.

And, finally, to what degree it is irrelevant. For the fact is that there are a great many behavioral problems that occur in mentally retarded people that are not contained in the DSM, unless they are fit into wastebasket categories like “organic personality disorder” or “atypical psychosis.” The only mental retardation categories that the DSM contains are the old American Association for Mental Deficiency (AAMD) ratings, “mild,” “moderate,” “severe,” and “profound,” a slender offering, to be sure.
The Psychiatric Treatment Manual (PTM), the companion to the DSM, recognizes the limitations of established psychiatric categories in retarded people (Gualtieri, 1989a). As far as treatment is concerned, the PTM has taken this approach: there is a role for the traditional psychiatric categories, albeit a small one. There is also a role for an alternative diagnostic schema.

The alternative schema for diagnosis includes a body of neurobehavioral disorders that are classified as specific mental retardation syndromes, like Lesch–Nyhan and Down’s. These are referred to as “pathobehavioral syndromes” because they are associated with a predictable constellation of pathological behaviors (Lesch–Nyhan and self-injurious behavior), or with the development of a specific neuropsychiatric disorder (Down’s syndrome and Alzheimer’s disease).

The alternative schema also includes a group of disorders that do not fit into either the traditional psychiatric or the pathobehavioral categories. These disorders, defined in purely behavioral terms such as aggression, pica, self-injurious behavior, and stereotypy, are referred to as “behavioral disorders,” which is not a very imaginative term.

The alternative schema also includes a group of disorders like phenylketonuria (PKU) and the Prader–Willi syndrome that might be considered “pathobehavioral syndromes,” but which are joined under the hypothetical construct “disorders of serotonin regulation.” This is a purely theoretical construction that is presented for the sake of argument. There may prove to be many alternative approaches to neuropsychiatric diagnosis like this one as our knowledge of the chemistry of behavior advances.

The organization of this chapter, then, is around four categories of neuropsychiatric disorders: traditional psychiatric disorders, behavioral disorders, pathobehavioral syndromes, and hypothetical constructs based on the neurochemistry of behavior. The system has a certain organizational simplicity, at least for the purpose of composing a chapter. It also has a certain utility, in clinical terms, because it reflects the current state of neuropsychiatric practice in the field of mental retardation. It also reflects the interests of researchers in the field, who write, for example, about “The Diagnosis of Major Affective Disorder,” or “The Treatment of Self-Injurious Behavior,” or “The Neurobehavioral Correlates of William’s Syndrome,” and who will one day be writing about “Behavioral Manifestations of Serotonin Dysregulation.”

It is, however, a purely arbitrary division. It is simple, but it is by no means pure. There is substantial overlap among the categories. The syndrome of Cornelia de Lange (CDLS), for example, is a pathobehavioral syndrome. But the prevailing problems of CDLS patients are best expressed in behavioral terms: self-injurious behavior, aggression, hyperactivity. And the most interesting question to raise about CDLS is whether there is an association to the major affective disorders, especially that class of the affective disorders that is linked to an abnormality of serotonin metabolism.

So, we do not propose a theory of how the abnormal behaviors of mentally retarded people may be classified. We only propose the simple idea that alternative models, or scheme, are necessary to embrace the full range of abnormal behaviors.