JBRF Expert Diagnostic Workshop

Definitions of Proposed Phenotypes

Narrow Phenotype (elated mood)

This phenotype is characterized by abnormal elevated mood states. A distinct episode of hypomania or mania of at least four days duration is required, during which time the child had elevated, elated mood or grandiosity. In addition, at least three other DSM-IV B symptom criteria must be met for diagnosis. In the absence of elated mood, three symptom criteria must be met in addition to grandiosity.

Symptom criteria include all 7 DSM-IV symptom criteria: inflated self-esteem or grandiosity, decreased need for sleep, more talkative than usual or pressure to keep talking, flight of ideas or subjective experience that thoughts are racing, increase in goal-directed activity or psychomotor agitation, and excessive involvement in pleasurable activities that have a high potential for painful consequences. If the child also meets criteria for ADHD, the symptom criteria of distractibility and psychomotor agitation count toward the diagnosis of mania only if in excess of the child’s usual ADHD.

Broad Phenotype (angry, irritable mood, chronic hyperarousal, explosiveness)

This phenotype is designed to capture children who have chronic irritability and hyperarousal. It is also called severe mood and behavioral dysregulation, and it differs from the narrow phenotype in that the child has no euphoria or grandiosity and no discernable episodes.

All of the following are required: 1) Chronic explosiveness, i.e., the child exhibits marked reactivity to negative emotional stimuli at least 3 times a week, on average. For example, the child’s response to frustration is associated with extended temper tantrums, verbal rages, and/or displays of aggression toward people or property; 2) Baseline abnormal mood (i.e., even between outbursts the child is angry or irritable); and 3) Chronic hyperarousal. To meet this last criterion, the child must have three of the following symptoms: distractibility, racing thoughts or flight of ideas, pressured speech, intrusiveness, agitation, and insomnia, nearly every day. In addition to the absence of elated mood or grandiosity, the presence of irritable mood distinguishes the broad phenotype from the narrow phenotype.

Core Phenotype (Episodic and abrupt transitions in mood state, and poor modulation of at least one drive state)

The hallmark features of this phenotype are episodic and abrupt transitions in mood state (mania/hypomania, depression, mixed state) and poor modulation of at least one drive (aggressive, sexual, appetitive, acquisitive). Descriptive definitions of mania/hypomania, depression, and mixed states remain essentially unchanged from DSM-IV. However, specifying daily, abrupt mood fluctuations and eliminating episode duration distinguish this phenotype from the others. These cardinal features must result in behaviors that are excessive or inappropriate for age and/or context and must be present on most days for at least 12 months to make the diagnosis.

Symptoms must not be due to the direct physiological effects of a substance or a general medical condition. In addition, the child must exhibit four or more of the following symptoms;
1) Excessive anger and oppositional and aggressive responses to situations that elicit frustration;

2) Poor self-esteem regulation (self-aggrandizement, exaggeration of abilities, and feelings of omnipotence, or, alternatively, pessimistic, self-critical, and overly sensitive to criticism or rejection);

3) **Sleep/wake cycle disturbances as exemplified by sleep discontinuity, sleep arousal disorders, or sleep/wake reversals;**

4) Excessive anxiety and fearfulness in response to novel or stressful situations;

5) A disturbance in the capacity to habituate to novel, loud or unexpected sounds and dissonant sensations;

6) Executive function deficits as exemplified by mental, emotional, or motor inflexibility;

7) A family history of recurrent mood disorder and/or alcoholism, as well as other bipolar spectrum disorders.