Using Phenotypic Expression in the Diagnosis and Treatment of Childhood Obesity

Ramona M. Wallace*, Alex L. Wallace

Muskegon Family Care, Muskegon, MI, USA

Abstract

Four phenotypes categorize patients participating in the Wellness Improvement Initiative program. I. Immediate BMI (Body Mass Index) Reducers expressed a readiness for change and immediately followed through with the nutrition and exercise portions of the program. II. Roadblocked BMI Reducers had physical issues that were uncontrolled, poorly controlled or undiagnosed. III. Psychological BMI Flat-liners had behavioral health issues, including depression and anxiety. IV. Roadblocked with Psychological Problems BMI Reducers experienced physical, psychological and social challenges. A systematic method of discovery, including these phenotypes, was used to individualize treatment. The greatest BMI reduction occurred with compliant participants in phenotypes I and II. The least successful participants faced a range of physical, psychological and social problems that likely affected compliance and certainly hampered weight control efforts.

Introduction

Obesity in children was once viewed as an exception. Now this disease state is so common that clinicians consider it the new normal and being overweight as typical [1]. Accurate and helpful educational resources about healthful eating and losing weight have become overwhelming for patients, due to aggressive marketing by the food industry. Additionally, clinicians must walk the fine line between invoking the social stigma of obesity and maintaining a productive patient/family relationship that acknowledges the significant negative health and cost associated with being significantly overweight. In 2010, the Muskegon Family Clinic instituted the Wellness Improvement Initiative to apply “readiness for change” principles from alcohol abuse studies to a childhood obesity pilot program [2].

During 24 months of the pilot study, patients who demonstrated readiness for change reduced overall Body Mass Index (BMI) through personalized interventions. Several trends emerged and they allowed categorization into four critical phenotypes of obese patients. Our intervention program now incorporates these phenotypes to provide personalized treatment that acknowledges individual barriers to change, as well as reinforcing personal attributes that facilitate weight loss.

Childhood Obesity Program: Wellness Improvement Initiative (WII)

The Childhood Obesity Program, Wellness Improvement Initiative (WII) operates at a Federally Qualified Health Center in inner city Muskegon, Michigan, which provides primary medical care to a highly underserved, multi-ethnic population. Almost 75% of the adult patient population (Figure 1) and 29% of the pediatric population is overweight or obese. The high prevalence of adult obesity presents a significant barrier to enticing children into healthy lifestyle instruction.

The WII program starts when a health care provider identifies an at-risk or obese child and refers children and their caretakers for evaluation by an obesity specialist, which includes a readiness-for-change assessment (see Appendix). Please note that it is vital to assess readiness for change in both the child and primary caretaker in order to create a social atmosphere of change and behavior modification. Together they are asked questions directed at recognizing, understanding and reflecting on the child’s weight problem [3]. Once they express a readiness for change, then a full history and physical examination is undertaken, with special consideration to their phenotype. We have identified four phenotypes (Figure 2).

I. Immediate BMI Reducers

This subset of children and their caretakers expressed a readiness for change immediately and followed through with the nutrition and exercise portions of the program. The children achieved an immediate and sustained reduction in the BMI for their age (Figure 3). They did not have any significant physical or emotional barriers for adopting the “5210” model as their behavioral modification program. This model stipulates 5 servings of fruit or vegetables, a limit of 2 hours of screen time, 1 hour of physical exercise, and 0 or no juice, pop or sugar-flavored drinks a day, as recommended by the Center for Disease Control [4].

II. Roadblocked BMI Reducers

The second subset of children had physical problems that were uncontrolled, poorly controlled or undiagnosed. When the

*Corresponding author: Ramona M. Kwapiszewski, DO, 2082 Geneva Lane, Norton Shores, MI 49441, USA, Tel: 517-256-8298; Email: drrkwap@gmail.com
Figure 1: Body Mass Index (BMI) for Adults at Muskegon Family Care. Caucasian assignment was based on stored values in the electronic medical records which were self-reported by patients on their first visit.

Figure 2: Percentage of Children (n=126) in Each of the Four Phenotypes of the WII Program.

Figure 3: Average Changes in BMI by Phenotype for Compliant and Noncompliant WII Program Participants. Noncompliance was determined by assessing food diaries, exercise regimens at home, and attendance at WII classes.
physical problems were addressed and treated, the child and caretakers were able to comply with the behavior modification and could implement the changes required for reducing BMI. Obstructive sleep apnea was the most common comorbidity or roadblock, followed by mood disorder, uncontrolled asthma, orthopedic and rheumatologic problems. Sleep disorders were assessed using a modified Epworth scale and a Mallampati score based upon the physical exam findings. If either measure demonstrated moderate risk, the children were sent for overnight oximetry monitoring. A full sleep study was ordered for high-risk children. Often, children after undergoing a tonsillectomy and an adenoidectomy when indicated, the weight reduction were significantly easier as long as they were compliant with the behavior modification. Asthma was addressed by first correctly identifying and adequately controlling the condition in children who were ready for change. After doing this, the children displayed a remarkable change in drive and were able to reduce their BMI rapidly.

The other remarkable subclasses of this roadblock phenotype included acanthosis nigricans, truncal obesity and increased neck diameter. A blood sample for ApoE genotype testing was drawn to help assess cardiac/metabolic risks and determine appropriate medication. An E3/3 genotype indicates a low risk for cardiovascular disease as an adult [5].

III. Psychological BMI Flat-liners

The third phenotype subset included children with behavioral health issues. Children with mood disorders who were taking an atypical antipsychotic drug typically gained weight or had a significantly harder time losing the weight if coping mechanisms led them to eating and inactivity.

Depression and anxiety were associated with disordered eating and often appeared as a coping mechanism. Sentinel events that triggered the behavioral problems, such as a father who was sent to prison, custody changes, primary caregiver changes and changes in environmental demographics, all contributed to disordered eating. Children described eating out of boredom, stress, loneliness and for comfort. Although attention deficit disorder could be considered a contributor to obesity, it was not included in this phenotype because of the number of children with this diagnosis who were taking a stimulant and then predictably lost weight.

IV. Roadblocked with Psychological Problems BMI Flat-liners

This phenotype included patients who were both roadblocked and experiencing behavioral health issues, an especially challenging combination. As might be expected, this phenotype was associated with scant success in reducing BMI (Figure 3), even among compliant participants.

V. Other Subgroups

Our pilot study revealed that there were functional subclasses
for the non-compliant patients and families just as there were for those who were ready to change (Figure 4). Among the non-compliant, targeted approaches can still improve their readiness to change, health outcomes and quality of life. The most dominant of the non-compliant functional classes included the “Uncontrolled Enablers”. In this class a caregiver, oftentimes a parent or grandparent, did not desire to make lifestyle changes conducive to improving the child’s health. The enabler prevented and sometimes reversed the progress that the child had made with the primary caregiver who was willing to change.

Another subgroup of patients who signed up for the WII program faced extenuating circumstances beyond their control (Figure 4B), which undercut compliance.

The Continuing Challenges of Identifying and Treating Obesity

In our pilot study, we noted that providers had to accept the challenge of identifying overweight/obese children during regular health care visits. Measuring BMI part of each visit can be successful in classifying overweight/obese children (Figure 5A). However, we also found that the majority of children in those categories did not have a recorded diagnosis of overweight/obesity (ICD-9 278; Figure 5B).

We also learned that providers typically avoid addressing obesity if the patient and family do not seem ready for change. Thus, the first and most significant barrier a patient may face is not hearing about their high-risk behaviors from providers. Our pilot program demonstrated that there were more patients in the ready-for-change group than in the not ready-for-change group, even though only 27% of eligible patients signed up for WII (Figure 4A). This is especially encouraging because ready-to-change patients can often identify their own barriers to weight reduction. Yet without a push from providers, patients may not initiate lifestyle changes. We encourage providers to be less hesitant to speak with overweight/obese patients and determine if they have any desire to improve their health choices. A frank discussion will identify the patients who have a good chance of reducing their BMI and staying within healthy constraints over time.

Another major barrier to provider intervention has been the poor outcome of intervention with patients/families that are not ready for change or are non-compliant. Thirty percent of eligible patients enrolled in the WII program (Figure 4A), and our non-compliant participants continued to gain weight (Figure 3B). Still, patients deserve the opportunity to change, even though they may not gain immediate advantage from it.

![Figure 5: Number of Children Ages 2 to 18 Who Were Obese or Overweight, Defined as BMI for Age (A) or Who Were Correctly Associated with an ICD-9 Diagnosis Code of 278 (B).](image-url)
Finally, our least successful participants (phenotypes III and IV) faced a range of psychological and social problems that likely affected compliance and certainly hampered weight control efforts. Additional support, including mental health intervention and social services, may ameliorate some of these problems.

**Discussion**

The pilot study uncovered phenotypes of overweight and obese children that can be easily identified and addressed to improve outcomes. Using the systematic method of discovery presented here to individualize treatment has been effective. Improving provider involvement in addressing obesity before it reaches morbidity remains an area for improvement in the healthcare community. Additionally, our study revealed the public health need to address the barriers of childhood obesity between the provider and patient, and to explore new ways to empower and prepare patients for change. Further community and peer support is also required to address childhood obesity, especially in the mental health arena. Provider education and follow through can enable assessment of the readiness for change by removing the social stigma of obesity. Lastly, developing tools for the four phenotypes would support clinicians in identifying barriers to weight loss.

**References**

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Appendix

1. THE ALGORITHM
   - Identify patient as overweight/obese (use BMI for age and growth charts available from the CDC)
2. Assess readiness for change (use the Patient Empowerment Model)
3. Assess barriers through history and physical examination (identify comorbidities, social and economic barriers and home lifestyle)
4. Identify patient phenotype
5. Address barriers according to phenotype (Treat comorbidities, address social and economic barriers)
6. Enroll and follow patient in nutrition and fitness program

   The behavioral changes that may work best may also be inappropriate for the patient and the patient's family. If this is the case, be sure to focus on positive behavior changes congruent with the family's value system, employing the values reflective of the patient’s social status, cultural beliefs and educational level. This will achieve better outcomes as patients and their families will be more receptive to the lifestyle changes and be able to maintain the changes longer.

INITIAL EVALUATION

1. History of Chief Complaint
   - Initial onset of obesity
   - duration
   - Amount of daily exercise
   - Number of meals a day at home
   - Number of meals a week at home
   - Amount of juice daily
   - Amount of soda daily
   - Amount of screen time a day
   - Amount of servings of fruit and green vegetables a day
2. Past Medical History
   - Orthopedic issues
   - Hypertension
   - Depression
   - Polycystic ovarian syndrome
   - Diabetes
   - Sleep apnea
   - Prader-Willi syndrome
   - Asthma
   - Acanthosis nigricans
3. Past Social History
4. Family History
   - Obesity
   - Asthma
   - Diabetes

   • Hypertension
   • Kidney disease

5. 5210 Scale
6. Readiness Scale
7. Depression Scale (center for Epidemiologic Studies Depression Scale for Children)
8. Lab Testing
   - BMI in 85th to 95th percentile with no risks: LIPID PROFILE
   - BMI in 85th to 95th percentile with risks: LIPID with CMP
   - BMI greater than 95th% percentile despite risks: LIPID with CMP
   - Asthmatics: allergy panel with CBC
   - Mallampati score of 3 or greater with obstructive sleep apnea symptoms: sleep study
9. Ongoing Evaluation and Monitoring
   - Every 3 months: height, weight, BMI, blood pressure
10. Assessment and Plan Using Diagnostic Coding
   - 278 obesity overweight
   - 278.01 obesity
   - 493 asthma
   - 327.23 obstructive sleep apnea syndrome
   - 401.1 hypertension
   - 272 hyperlipidemia
   - 311 depression

Schedule for fitness program at the YMCA, school, or facility.
Schedule monthly follow-up.
Institute asthma education, including spirometry and American Academy of Pediatrics materials.
Schedule sleep study/overnight oximetry labs.
Begin cognitive behavioral therapy.