

# Tyrosine measurement/ management in children with PKU: results of an International survey

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## INTRODUCTION

Executive Function (EF) deficits are prevalent in PKU, and are associated with an Attention Deficit Disorder diagnosis up to 5 times the norm.<sup>1</sup> EF deficits increase with higher levels of phenylalanine (phe).<sup>2</sup> Recent evidence implicates the dual role of high phe in combination with low tyrosine (tyr), expressed as the high phe:tyr ratio.<sup>3-5</sup> A high phe:tyr ratio is hypothesised to result in compromised dopamine synthesis, which contributes to EF deficits.<sup>3</sup> Phe:tyr monitoring/management may be useful in children with PKU,<sup>6</sup> however, practice among different metabolic clinics re tyrosine screening and/or treatment is largely unknown both within countries and internationally.

## METHOD

A short web-based survey was emailed to all metabolic clinics in Australia/New Zealand – 50% of clinics in this region responded. The same survey was then emailed to each National President/Secretary of the SSIEM for distribution. In total, 20 separate clinics from 12 countries completed the survey. 85% of surveys were completed by the clinic's consultant metabolic physician. The median number of children managed by each clinic was 60 (Mean = 105).

## PHE SCREENING + MANAGEMENT

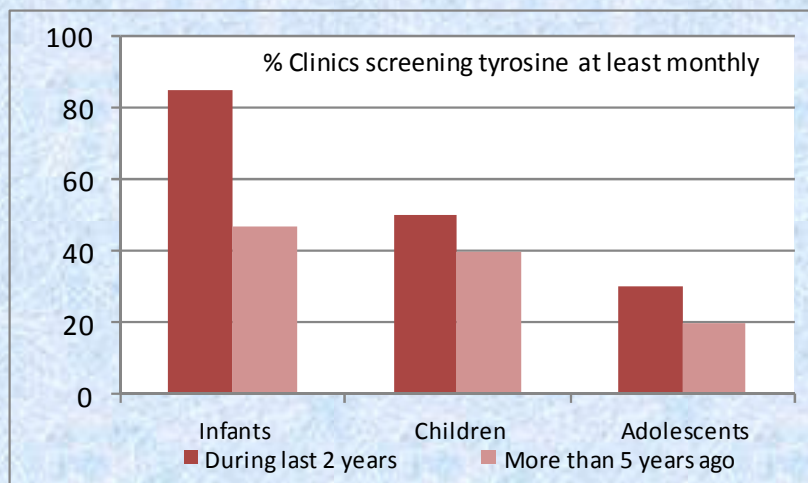
Frequency of phe screening was highly consistent across all clinics. 80% recommended infant phe screening every 1-2 weeks, dropping to monthly for adolescents. Attrition (non-adherence to recommended screening frequency) began during childhood (40%) and rose to 70% during adolescence. 50% of clinics used their country's guidelines as a reference point for phe recommendations

## EF MONITORING

50% of clinics routinely assessed children's cognitive /neuropsychological function.

## TYROSINE SCREENING + MANAGEMENT

Frequency of tyrosine screening was highly variable, both within countries and internationally. Tyrosine screening at least monthly has clearly been on the rise during the last 5 years with over 80% of clinics now routinely screening tyrosine levels in infancy.



25% of clinics supplemented some patients with tyrosine. Patients targeted for treatment were: those with tyrosine levels consistently below "normal"; adults with poor dietary control; pregnant women; & young children with well-controlled phe levels. No adverse events were reported. One clinic noted mood improvement *if* children had originally presented with depressive symptoms; another noted they rarely saw tyrosine deficiency if PKU formula and diet were followed. Dosage was usually the "least necessary" to shift tyrosine levels back to "normal"; 1 gram per day was also nominated as a practical starting point.

## DISCUSSION + THANK YOU

Practice in relation to phe screening was highly consistent. Tyr assessment and management practices were more variable. Trends towards increased screening are emerging; and of those clinics that responded, a quarter reported managing the tyr levels of at least some patients. These results may stimulate discussion regarding future tyr and EF screening and management practices.

Our sincere thanks to those clinics who volunteered data for this study, including sites located in Australia/NZ; USA; Canada; European nations & Asia. Your support of this important project is appreciated.

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