

2007 Advisory Committee Comments

| <i>Conditions</i> | <i>Affiliation</i> | <i>Name</i> | <i>Prevention</i> | <i>Treatment</i> | <i>Public Health</i> | <i>AvailableTechnology</i> | <i>Comments</i> |
|-------------------|--------------------|-------------------------------------|-------------------|------------------|----------------------|----------------------------|--|
| 3-MCC | | | | | | | |
| | | <i>Childrens Advocacy Groups</i> | Unsure | Yes | No | Yes | 1. Still some concerns regarding medical rationale. 2. Treatment is available if needed. |
| | | <i>Medical/Clinical Specialties</i> | Unsure | Yes | Unsure | Yes | 1. The clinical significance of 3-MCC def under dispute. 3. Unnecessary treatment can be possible. |
| | | | No | Unsure | No | Yes | 4. Only for C5OH. |
| | | <i>Parents</i> | Unsure | Yes | No | Unsure | 1. Insufficient datat available. 4. Technology seems insufficient to justify. |
| | | <i>Principle Payers</i> | No | Unsure | No | Yes | This is not a disease and given the medicalization intrinsic to our culture, has more potential to cause unnecessary worry than anything else. |
| | | <i>Professional Associations</i> | Unsure | Unsure | No | Unsure | 2. No consensus regarding optimal treatment. Medical experts need to evaluate individually to determine best course of action. |
| | | | No | Unsure | No | No | 2. If condition exists a (????????? Not ue ????????) appropriate (???? Rx/Px ????) |
| | | | No | Unsure | No | Yes | 2. Unclear whether treatment is needed. Concern - ethically must follow up. Interest in how to (learn/research?) outcomes. |

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|-------------------------------------|--------------------|-------------|-------------------|------------------|----------------------|----------------------------|--|
| BKT | | | | | | | |
| <i>Medical/Clinical Specialties</i> | | | | | | | |
| | | | Yes | Yes | Yes | Yes | 4. Will miss majority of cases, other than severe. |
| CbIA,B | | | | | | | |
| <i>Principle Payers</i> | | | | | | | |
| | | | Yes | Yes | Yes | Yes | Available Technology - Yes but (high or elevated) false positives. |
| | | | Unsure | Unsure | Yes | Unsure | For available technology both yes and unsure were indicated with this note: There is more uncertainty that treatment will eliminate neurological degeneration. |

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|-------------------|--------------------|-------------------------------------|-------------------|------------------|----------------------|----------------------------|--|
| CPT-1 | | | | | | | |
| | | <i>Childrens Advocacy Groups</i> | Yes | Yes | Unsure | Unsure | 1. Still seems that there is uncertainty, re: testing because of outcome with severe, mild? 2. TMS will not pick up all cases - seems only mild. |
| | | <i>Ethics/Public Health</i> | Yes | Yes | Unsure | Yes | 1. Incidence in WA is not clear. Still think that screen in WA is justified on a provisional basis. |
| | | <i>Medical/Clinical Specialties</i> | Yes | Yes | Yes | Yes | 1. Should consider mutation analysis on targeted population. |
| | | | Yes | Yes | Yes | Yes | Answer Yes in all four catagories for the Severe form. For mild form: Prevention - Unsure; Treatment - Unsure; Public Health - Yes; Technology - Unsure. |
| | | <i>Parents</i> | Yes | Yes | Yes | Yes | 4. Technology will at least pick up some cases though not perfect. Knowledge of specific ethnic groups will lead to further testing. |
| | | <i>Principle Payers</i> | Yes | Yes | Yes | Unsure | I'm not sure that the rationale for including this test as a region - specific screeni is met because of the lack of sensitivity of the test. |
| | | <i>Professional Associations</i> | Yes | Yes | Yes | Unsure | 1. Stating that this is tested, but many may be missed. |

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|-------------------|-------------------------------------|-------------------|-------------------|------------------|----------------------|----------------------------|--|
| CUD | | | | | | | |
| | <i>Childrens Advocacy Groups</i> | | Yes | Yes | Yes | Yes | 4. At least will identify some, sounds like most. |
| | <i>Medical/Clinical Specialties</i> | | | | | | |
| | | Judith Martin, MD | Yes | Yes | Unsure | Unsure | 3. Rare disorder but severe consequences. 4. Will not detect all and complex interpretation. |
| | | | Yes | Yes | Unsure | Unsure | 3. Difficulty with sensitivity and specificity. 4. Variation in infant age (premies) vs carnitine quantitation. |
| | <i>Parents</i> | | Yes | Yes | Yes | Yes | 4. Current technology will pick up some cases, though not perfect. |
| | <i>Professional Associations</i> | | Yes | Yes | Yes | Unsure | 4. False positives |
| HMG | | | | | | | |
| | | | Yes | Yes | Yes | Yes | 4. Will likely miss mild cases. |
| | <i>Ethics/Public Health</i> | | | | | | |
| | | Benjamin Wilfond, | No | Yes | No | Unsure | 4. Identifies a large number of conditions. |
| | <i>Medical/Clinical Specialties</i> | | Yes | Yes | Yes | Unsure | The public health rationale for screening is great! The screening test results are currently so non-specific that I'm not convinced the use of the test will perform a usefull/successful public health funcion. |
| | <i>Parents</i> | | Yes | Yes | Yes | Yes | 3. I believe the frequency rate and potential prevention of dire complications justify screening. |

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|-------------------------|--------------------|-------------|-------------------|------------------|----------------------|----------------------------|--|
| MUT | | | | | | | |
| <i>Principle Payers</i> | | | | | | | |
| | | | Yes | Unsure | Yes | Yes | Available Technology - yes with (high or elevated) false positives. |
| | | | Unsure | Unsure | | Unsure | Both Prevention Potential and Treatment had this note: Treatment with variable effectiveness, poor QOL very possible. For Available Technology both Yes and Unsure were checked with this note: Technology less sensitive. |
| PROP | | | | | | | |
| <i>Principle Payers</i> | | | | | | | |
| | | | Unsure | Unsure | Yes | Unsure | Treatment & screening are not great for this set of disorders and treatment frequently doesn't resolve symptomatology or prognosis. |
| | | | Yes | Unsure | Yes | Yes | Available Technology - (High or Increased) false positives |
| TYR-1 | | | | | | | |
| | | | Yes | Yes | Yes | No | 1. Succinylacetone test is likely the only way to detect Tyr-1. The sensitivity of the assay under investigation. |