Canadian HPP Clinical Expert Committee

APPLICATION & CLINICAL SUMMARY FORMS
for Hypophosphatasia (HPP) Enzyme Replacement Therapy

Please FAX all requests to AMY @ 204-789 3907 or scan and fax to ayakimoski@chrim.ca

Please ensure to include ALL RELEVANT REPORTS

REFERRING DOCTOR _____**Institution** _____

City _____Province/Territory _____

Email _____
Telephone_____

PATIENT INFORMATION

Initials/Case #: _____Date of Birth _____
Day/Month/Year

Province/Territory of Residence _____ Sex: □ Male □ Female

TYPE OF REQUEST □ Antenatal □ Newborn □ Infantile □ Juvenile (Childhood)

CRITERIA TO QUALIFY PATIENT FOR ERT (check all that apply)

Patient must have confirmed diagnosis of HPP #1 and fulfill criteria in 2 other areas as listed below in #2 - #6 (known to respond to ERT)

#1. □ **Confirmed diagnosis of Pediatric-Onset HPP**

#2. □ **Clinical manifestations (check all that apply):**

- □ Failure to Thrive/Poor Growth (crossing percentiles)
- □ Clinical signs of rickets: thickened wrists; rachitic rosary
- □ Fractures, recurrent and poorly healing
- □ Orthopedic Surgical Procedures
- □ Abnormal Gait
- □ Delayed Motor Development
- □ Use of Assistive Devices

#3. □ **Radiologic findings (check all that apply):**

- □ Radiologic signs of rickets
- □ Severe hypomineralization/Osteopenia
- □ Limb Bowing/Deformation
- □ Other eg radiolucent tongues_____

#4. □ **Severe Pain plus #1 <u>and</u> #2 or #3**

□ Pain that is not related to a fixable problem (eg, pain that is not self-limited due to a surgical procedure or fracture) AND

□ Pain severe enough to repeatedly miss school days

□ Pain requiring opioid or non-opioid analgesics on a daily basis

#5. □ **Respiratory Findings (check all that apply):**

□ Lung hypoplasia
□ Respiratory Failure
□ Supplemental Oxygen
□ Assisted Ventilation

#6. □ For **neonatal requests for perinatal HPP**, requesting centre must have management team & decision making structure in place and follow guideline for initiation & monitoring of treatment of neonate with perinatal HPP (Attachment 1).

For HPP CEC Notes:

1. Application received _____
2. Application disseminated to committee members _____
3. Recommendations received from committee members and collated _____
4. Conference call, if necessary _____
5. Committee recommendation_____ submitted _____

HPP CLINICAL SUMMARY FORM –Please Fax with application form & all relevant reports

SYSTEM	FEATURE/CRITERIA **BOLD** items are those criteria that can qualify patient for ERT (respond to ERT) Items not in bold are features of HPP *not known* to respond to ERT
Growth	Current height _____ cm feet/inches (circle one) (_____%) Current weight _____ kg pounds (circle one) (_____%) Current Head circumference _____ cm (_____%) ☐ **Poor growth (crossing percentiles)/FTT** ☐ **Yes** ☐ No
Genetic studies	☐ **Confirmed diagnosis of Pediatric-Onset HPP** ☐ Low ALP, specify:_____ ☐ *ALPL* mutation, specify:_____
Clinical Manifestations	☐ **Fractures** **Number**_____ **Location**_____ ☐ **Non-healing** ☐ **Healed** ☐ **Thickened wrists ☐ yes ☐ no; rachitic rosary ☐ yes ☐ no** ☐ **Orthopedic surgical procedures** **Specify type of surgery, age at procedure:** _____ ☐ **Gait** ☐ Normal ☐ **Abnormal, specify** _____ ☐ **Motor development:** ☐ Meeting age-appropriate milestones ☐ **Delayed** ☐ **Weakness**, specify _____ **Details:**_____ _____ ☐ **6 Minute Walk Test** ☐ **Yes (attach result)** ☐ No ☐ **Use of assistive devices:** ☐ **Yes, specify**_____ ☐ No ☐ Scoliosis ☐ Skeletal deformities, specify_____ Details:_____ _____ ☐ Craniosynostosis ☐ Early loss of primary teeth. Age at first tooth loss: _____Months/Years (Circle One) ☐ Poor dentition ☐ Other teeth problems:_____ ☐ Joint swelling, specify_____ ☐ Participates in sports: ☐ Yes ☐ No ☐ Not applicable ☐ Misses school: ☐ Yes ☐ No ☐ Not applicable ☐ Other eg Childhood Health Assessment Questionnaire (CHAQ) Disability Index Details:_____

X-ray findings and other Imaging Results	□ **Rickets** □ **Severe hypomineralization/osteopenia** □ **Limb Bowing/Deformation** □ **Other, please specify: eg radiolucent tongues**_____ □ **Skeletal survey**□ Yes □No If yes, age at x-rays _____Months/Years (Circle One) X-ray findings_____ Most recent X-ray results_____ □ DEXA Scan □ Yes □ No If yes age(s) at DEXA_____Months/Years (Circle One) Results_____ □ Peripheral quantitative computed tomography (pQCT) (research tool)
Pain	□ **Pain that is not related to a fixable problem** (ie: pain that is not self-limited d/t a surgical procedure or fracture) □ **Pain severe enough to repeatedly miss school days** □ **Daily use of opioid or non-opioid analgesics** □ Analgesics (overall use) □ Yes □ No List analgesics used, dosage, frequency:_____ _____ □ **Muscle pain** □ **Yes** □ **No** □ **Bone pain** □ **Yes** □ **No** □ **Joint pain** □ **Yes** □ **No** Specify type of pain, location, pain at rest or with activity, daytime or at night, and clinical response to intervention _____ □ Heating Pad □ Yes □ No □ Massage □ Yes □ No □ Other interventions, specify: _____ □ Validated pain tools_____ eg. Visual analog for pain eg**.** Childhood Health Assessment Questionnaire (CHAQ) Pain Index eg**. "**PROMIS" http://www.healthmeasures.net/explore-measurement-systems/promis □ Patient's/parent's assessment of pain and QOL
Respiratory	□ **Lung hypoplasia** □ **Yes** □No □ **Respiratory failure** □ **Yes** □ No □ **Supplemental O2** □ **Yes** □ No □ **Assisted ventilation** □ **Yes, specify**_____ □ No
Renal Manifestation	□ Nephrocalcinosis □ Yes □ No □ Renal failure/reduced renal function □ Yes □ No
Other studies	□ Hearing Loss □ Yes if Yes, specify_____ □ No □Not tested □Seizures □ Yes If Yes, B6 responsive □ Yes □No □ No □ Cognitive development □ Normal for age □ Delayed, if delayed, specify: _____ _____
Biochemical parameters	□ Calcium, phosphate, magnesium, **alkaline phosphatase**, PTH, 25 OH vitamin D, **pyridoxal-5-phosphate (PLP)**, urine phosphoethanolamine (PEA)