

Hypophosphatemic Rickets

Hypophosphatemic rickets is a genetic condition in which the kidneys are unable to retain phosphorus within the body, which results in the loss of excessive amounts of phosphorus into the urine. Consequently, there is not enough phosphorus remaining in the bloodstream to build healthy bones, and the affected child develops rickets. There is also a defect in converting vitamin D to its activated form.

How is hypophosphatemic rickets diagnosed?

The diagnosis of hypophosphatemic rickets is usually made when an infant or toddler presents with rickets (see the handout *Vitamin D Deficiency and Rickets*). Further testing shows a normal blood calcium level, a very low blood phosphorus level, and an elevated urinary phosphorus level. The 25-hydroxy-vitamin D level is normal, but the 1,25-dihydroxy-vitamin D level is low. X-rays will show the evidence of rickets in the bones, similar to that seen with vitamin D deficiency. In some cases, the diagnosis of hypophosphatemic rickets is made because another family member is similarly affected. Genetic testing is sometimes available for this condition.

How is hypophosphatemic rickets treated?

The treatment for hypophosphatemic rickets is phosphate replacement. The phosphate dosage must be given at least 4 times a day, generally with each meal and at bedtime, to provide the body with a steady supply of phosphorus for the bones. Most patients with hypophosphatemic rickets also require a dosage of calcitriol (Rocaltrol®) or

alfacalcidol (One-Alpha®) to help absorb the calcium and phosphorus into the bones and to heal the rickets.

What are phosphate supplements?

Phosphate supplements increase the phosphate level in the blood. Two main forms of are:

- JAMP-Sodium Phosphate: a large effervescent tablet (500 mg)
- Phoslax®: a clear liquid (125 mg/mL)

How should you give your child phosphate supplements?

JAMP-Sodium Phosphate: dissolve prescribed amount in at least 250 mL (1 cup) of juice or water (**not milk!**). Divide this into 4 doses per day.

Phoslax®: Measure the prescribed amount in a syringe. Mix the dose with at least an equal amount of water or juice (**not milk!**) and have your child drink this quickly, or have your child drink the prescribed amount quickly and follow with a drink of juice or water.

What should you do if your child misses a dose?

Your child can take the missed dose as soon as you remember. If it is almost time for the next dose, skip the missed dose. Do not give your child two doses together to make up the missed dose.

What are the possible side effects of phosphate supplements?

Your child may have some side effects when taking phosphate supplements. Let your

doctor know if the side effects don't go away or if they bother your child.

- stomach upset
- dizziness
- headache

Call your child's doctor right away or take your child to the nearest Emergency Department if they have any of these side effects.

- severe skin rash or hives
- trouble breathing (wheezing or shortness of breath)
- swelling of face, lips, tongue, or throat
- unusual tiredness or weakness
- severe vomiting or watery diarrhea
- severe dizziness

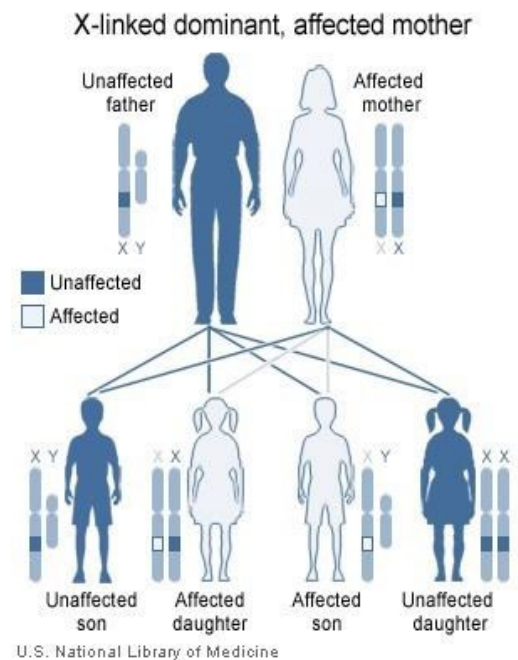
What other important information should you know about phosphate supplements?

- Phosphate supplements can interfere with how the body absorbs some other medicines; therefore, your child should take them at least two hours before or after antacids and calcium, magnesium or iron supplements.
- Keep phosphate supplements at room temperature in a cool, dry place away from sunlight. Do not store them in the bathroom, as heat and steam can damage them.

Is hypophosphatemic rickets hereditary?

In many cases, hypophosphatemic rickets has what is known as X-linked dominant inheritance. Affected mothers pass the conditions to 50% of their daughters and sons; their other children are unaffected. Affected fathers pass the condition to all of their daughters, but not to their sons. Males are generally more affected than females.

Other patterns of inheritance are possible. If this is suspected, your doctor can refer your family to a Medical Geneticist for further evaluation and counselling.



Questions from Families

Q: Can I use non-prescription vitamin D instead of calcitriol (Rocaltrol®) or alfacalcidol (One-Alpha®)?

A: *No! Vitamin D from the drugstore doesn't work in the body until it is turned into the activated form of vitamin D. Because of his or her medical condition, your child's body is not able to convert vitamin D into the activated form.*

Q: My son is booked for a surgical procedure soon. I've been told he should have nothing to eat or drink before the surgery. Does this include the phosphorus and calcitriol (Rocaltrol®) or alfacalcidol (One-Alpha®)?

A: *Speak with your endocrinologist about this— it can be dangerous to miss any doses. If the calcium level drops too low, the surgery will be cancelled. Often the doctor recommends taking all doses of medication with just a tiny sip of water.*

Q: What should I do if my child is vomiting and can't keep his medicine down?

A: *Since it can be dangerous to miss doses, you should speak to your endocrinologist about this.*

Q: I don't like giving my child medicine. Can I give her a special diet instead of the phosphorus and Rocaltrol® or One-Alpha®?

A: *Every day a child's body needs phosphorus. Unfortunately, it is very difficult for your child to get enough phosphorus from diet alone, so she needs supplements as well. One of the activated forms of vitamin D, either calcitriol (Rocaltrol®) or alfacalcidol (One-Alpha®), is also needed in order for the calcium and phosphorus from the diet and from supplements to be absorbed by the digestive system.*

As you can see, the process of treating your growing child with a disorder of calcium or phosphorus involves taking medications regularly and checking blood and urine to be sure the amounts are right for her. At first, it will take a lot of care, but soon it will become part of your everyday life, and you will be able to enjoy your child for the unique child that he or she is.

Websites and support groups for disorders of calcium and phosphorus

X-Linked Hypophosphatemia Network:
<http://www.xlhnetwork.org>

HealthLink BC: Food Sources of Calcium and Vitamin D:
<https://www.healthlinkbc.ca/healthlinkbc-files/sources-calcium-vitamin-d>

Medic Alert Canada:
<http://www.medicalert.ca>

More links are available from the **BC Children's Hospital Endocrinology & Diabetes Unit:**
<http://endodiab.bcchildrens.ca>

Appendix

Selected Canadian Calcium Products	
Generic Name / Brand Name	Elemental Calcium
Tums® Regular 500 mg	200 mg/tab
Tums® Extra Strength 750 mg	300 mg/tab
Tums® Ultra Strength 1000 mg	400 mg/tab
Viactiv® Chews	500 mg/chew
BCCH Pharmacy suspension	80 mg/mL
<p>Note: The regular Tums® tablet, for example, is called Tums® 500 mg. Since calcium carbonate is 40% elemental calcium, Tums® 500 mg actually contains only 200 mg of elemental calcium.</p>	

Canadian Vitamin D Products	
Generic Name	Trade Name and Dosages Available
Cholecalciferol (vitamin D ₃)	Multivitamins: most contain 400 IU Supplements: usually 400 IU or 1000 IU <ul style="list-style-type: none"> • Baby Ddrops®: 400 IU/drop • Kids Ddrops®: 400 IU/drop • Adult Ddrops®: 1000 IU/drop
Alfacalcidol (1-hydroxy-vitamin D)	One-Alpha®: <ul style="list-style-type: none"> • 0.25-microgram capsules • 1-microgram capsules • 2 microgram/mL (0.1 microgram/drop)
Calcitriol (1,25-dihydroxy-vitamin D)	Rocaltrol®: <ul style="list-style-type: none"> • 0.25-microgram capsules • 0.5-microgram capsules

Normal Levels of Common Lab Tests for a Child 6–12 Months of Age*	
Test	Normal Range
calcium*	1.87–2.50 mmol/L
ionized calcium*	1.10–1.30 mmol/L
phosphorus*	1.29–2.58 mmol/L
magnesium*	0.78–1.03 mmol/L
intact PTH	1.0–5.5 pmol/L
alkaline phosphatase	110–320 U/L
25-hydroxy-vitamin D	25–110 nmol/L
1,25-dihydroxy-vitamin D	40–190 nmol/L
urinary calcium/creatinine ratio*	<1.69 mmol/mmol
<p>*Normal levels vary depending on the age of the child and the lab method used.</p>	

Canadian Phosphorus Products	
Generic Name	Trade Name and Dosage Available
Sodium phosphate monobasic	JAMP-Sodium Phosphate fizzy tablets: 500 mg or 16.1 mmol elemental phosphorus per tab
Sodium phosphate monobasic, dibasic	Phoslax® oral solution, 45-mL bottle: 129 mg or 4.15 mmol elemental phosphorus per mL
Potassium phosphate monobasic (Health Canada Special Access Programme)	K-Phos® Original 500-mg tabs: 114 mg or 3.68 mmol elemental phosphorus per tab
Potassium phosphate monobasic, dibasic	Potassium Phosphates Injection USP, 50-mL vial: 93 mg or 3.0 mmol elemental phosphorus per mL