Supplementary figure 1 Recruitment of patients with hereditary hypophosphatemia

Diagnosis E83.3 Patients identified by contacting hospital departments N = 29

Diagnosis E83.3 Additional patients identified through Norwegian Patient Register search N = 9

Excluded Secondary hypophosphatemia N = 5 Hypophosphatasia N = 4

Eligible Hereditary hypophosphatemia N = 29

Not included - No response to repeated invitation N = 1

Included N = 28 (18 female, 10 male)

PHEX mutation N = 21
FGF23 mutation N = 0
DMP1 mutation N = 0
ENPP1 mutation N = 0
FAM20C mutation N = 2
SLC34A3 mutation N = 1
No mutation identified N = 4
Exclusion criteria:
- Primary hyperparathyroidism is characterized by elevated or high normal levels of serum intact parathyroid hormone combined with serum calcium above upper normal reference range.
- Secondary hyperparathyroidism in renal failure or malabsorption
- Tertiary hypoparathyroidism is diagnosed in patients with secondary hyperparathyroidism, when elevated levels of serum intact PTH and serum calcium persist upon withdrawal of the stimulus for secondary hyperparathyroidism.
- Vitamin D dependent rickets types I and II are characterized by hypocalcemia combined with low 1,25 (OH)2 vitamin D; hypocalcemia is not a feature of hypophosphatemic rickets.
- Fanconi syndrome or other tubulopathy,
- Vitamin D deficiency

Hypophosphatemia secondary to acute metabolic derangements.