

**eMERGE  
Northwestern University  
Height phenotype algorithm**

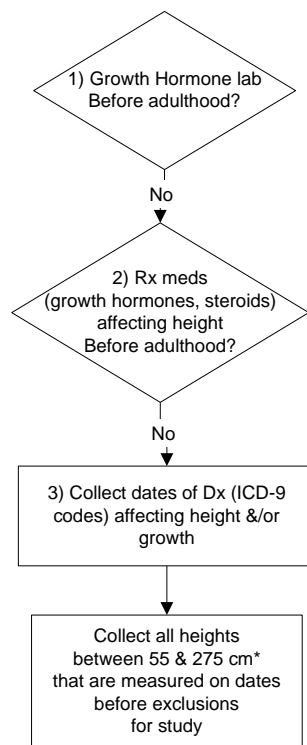
Below is a flowchart used to select patients and their height measures for a GWAS (Genome Wide Association Study) from an EMR (Electronic Medical Record). Lists of diagnoses, labs, and medications referred to in the flowchart can be found on the following pages.

If you have questions regarding this algorithm, you may contact either:

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**Flowchart:**



\*lowest & highest measured height for adults on record anywhere in the world

**APPENDIX of Medications and Diagnoses**

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**1) Growth Hormone lab during childhood** (females <= age 18, males <= age 21) (if lab ordered during childhood exclude patient altogether):

LOINC code 2963-7 :

“growth hormone” or “human growth hormone”

**2) Medications during childhood** (females <= age 18, males <= age 21) (if prescribed during childhood exclude patient altogether):

Generic name	Pharmaceutical class name	Source name	Source type	Source ID
Aminoglutethimide	Adrenal Steroid Inhibitors	RXNORM	IN (Ingredient)	677
trilostane	Adrenal Steroid Inhibitors	RXNORM	IN	38668
Oxymetholone	Anabolic Steroids	RXNORM	IN	7813
Oxandrolone	Anabolic Steroids	RXNORM	IN	7779
Stanozolol	Anabolic Steroids	RXNORM	IN	10032
Nandrolone	Anabolic Steroids	RXNORM	IN	7244
testosterone (inc. topical)	androgens and anabolic steroids	RXNORM	IN	10379
pegvisomant	Growth Hormone Receptor Antagonists	RXNORM	IN	278739
Sermorelin	Growth Hormone Releasing Hormones (GHRH)	RXNORM	IN	56188
somatrem	Growth Hormones	RXNORM	IN	56570
Somatropin	Growth Hormones	RXNORM	IN	61148

### **3) ICD-9 Dx affecting height:**

**Disorders in childhood (females <= age 18, males <= age 21) that would affect height (if occurred during childhood exclude patient altogether)**

#### **Tuberculosis or echinococcus of thyroid or adrenal glands**

017.5	Tuberculosis of thyroid gland
017.50	Tuberculosis of thyroid gland, confirmation unspecified
017.51	Tuberculosis of thyroid gland, bacteriological or histological examination not done
017.52	Tuberculosis of thyroid gland, bacteriological or histological examination unknown (at present)
017.53	Tuberculosis of thyroid gland, tubercle bacilli found (in sputum) by microscopy
017.54	Tuberculosis of thyroid gland, tubercle bacilli not found (in sputum) by microscopy, but found by bacterial culture
017.55	Tuberculosis of thyroid gland, tubercle bacilli not found by bacteriological examination, but tuberculosis confirmed histologically
017.56	Tuberculosis of thyroid gland, tubercle bacilli not found by bacteriological or histological examination, but tuberculosis confirmed by other methods (inoculation of animals)
017.6	Tuberculosis of adrenal glands
017.60	Tuberculosis of adrenal glands, confirmation unspecified
017.61	Tuberculosis of adrenal glands, bacteriological or histological examination not done
017.62	Tuberculosis of adrenal glands, bacteriological or histological examination unknown (at present)
017.63	Tuberculosis of adrenal glands, tubercle bacilli found (in sputum) by microscopy
017.64	Tuberculosis of adrenal glands, tubercle bacilli not found (in sputum) by microscopy, but found by bacterial culture
017.65	Tuberculosis of adrenal glands, tubercle bacilli not found by bacteriological examination, but tuberculosis confirmed histologically
017.66	Tuberculosis of adrenal glands, tubercle bacilli not found by bacteriological or histological examination, but tuberculosis confirmed by other methods (inoculation of animals)
122.2	Echinococcus granulosus infection of thyroid

#### **Benign or malignant neoplasm of thyroid, parathyroid, adrenal, or pituitary glands**

193	Malignant neoplasm of thyroid gland
194.0	Malignant neoplasm of adrenal gland
194.1	Malignant neoplasm of parathyroid gland
194.3	Malignant neoplasm of pituitary gland and craniopharyngeal duct
198.7	Secondary malignant neoplasm of adrenal gland
226	Benign neoplasm of thyroid glands
227.0	Benign neoplasm of adrenal gland
227.1	Benign neoplasm of parathyroid gland
227.3	Benign neoplasm of pituitary gland and craniopharyngeal duct (pouch)
237.0	Neoplasm of uncertain behavior of pituitary gland and craniopharyngeal duct
237.2	Neoplasm of uncertain behavior of adrenal gland
242.4	Thyrotoxicosis from ectopic thyroid nodule

242.40	Thyrotoxicosis from ectopic thyroid nodule without mention of thyrotoxic crisis or storm
242.41	Thyrotoxicosis from ectopic thyroid nodule with mention of thyrotoxic crisis or storm
V10.87	Personal history of malignant neoplasm of thyroid

### **Hypo- or hyperactivity of thyroid, parathyroid, adrenal or pituitary glands**

244	Acquired hypothyroidism
244.0	Postsurgical hypothyroidism
244.1	Other postablative hypothyroidism
244.2	Iodine hypothyroidism
244.3	Other iatrogenic hypothyroidism
244.8	Other specified acquired hypothyroidism
244.9	Unspecified hypothyroidism
245	Thyroiditis
245.0	Acute thyroiditis
245.1	Subacute thyroiditis
245.2	Chronic lymphocytic thyroiditis
245.3	Chronic fibrous thyroiditis
245.4	Iatrogenic thyroiditis
245.8	Other and unspecified chronic thyroiditis
245.9	Unspecified thyroiditis
246	Other disorders of thyroid
246.2	Cyst of thyroid
246.3	Hemorrhage and infarction of thyroid
246.8	Other specified disorders of thyroid
246.9	Unspecified disorder of thyroid
252	Disorders of parathyroid gland
252.0	Hyperparathyroidism
252.00	Hyperparathyroidism, unspecified
252.01	Primary hyperparathyroidism
252.02	Secondary hyperparathyroidism, non-renal
252.08	Other hyperparathyroidism
252.1	Hypoparathyroidism
252.8	Other specified disorders of parathyroid gland
252.9	Unspecified disorder of parathyroid gland
253	Disorders of the pituitary gland and its hypothalamic control
253.1	Other and unspecified anterior pituitary hyperfunction
253.4	Other anterior pituitary disorders
253.7	Iatrogenic pituitary disorders
253.8	Other disorders of the pituitary and other syndromes of diencephalohypophyseal origin
253.9	Unspecified disorder of the pituitary gland and its hypothalamic control
255	Disorders of adrenal glands
255.3	Other corticoadrenal overactivity
255.4	Corticoadrenal insufficiency
255.5	Other adrenal hypofunction

255.6	Medulloadrenal hyperfunction
255.8	Other specified disorders of adrenal glands
255.9	Unspecified disorder of adrenal glands
263.2	Arrested development following protein-calorie malnutrition
270.0	Disturbances of amino-acid transport
277.5	Mucopolysaccharidosis
377.51	Disorders of optic chiasm associated with pituitary neoplasms and disorders
577.8	Other specified disease of pancreas
588.81	Secondary hyperparathyroidism (of renal origin)
628.1	Female infertility of pituitary-hypothalamic origin
648.1	Thyroid dysfunction complicating pregnancy, childbirth, or the puerperium
648.10	Thyroid dysfunction of mother, complicating pregnancy, childbirth, or the puerperium, unspecified as to episode of care
648.11	Thyroid dysfunction of mother, with delivery
648.12	Thyroid dysfunction of mother, with delivery, with mention of postpartum complication
648.13	Thyroid dysfunction, antepartum
648.14	Thyroid dysfunction, postpartum
790.94	Euthyroid sick syndrome
794.5	Nonspecific abnormal results of thyroid function study

#### **Injury to thyroid gland**

848.2	Sprain and strain of thyroid region
868.01	Adrenal gland injury without mention of open wound into cavity
868.11	Adrenal gland injury, with open wound into cavity
874.2	Open wound of thyroid gland, without mention of complication
874.3	Open wound of thyroid gland, complicated

#### **Exogenous hormone**

962.0	Poisoning by adrenal cortical steroids
962.4	Poisoning by anterior pituitary hormones
962.5	Poisoning by posterior pituitary hormones
962.6	Poisoning by parathyroid and parathyroid derivatives
962.7	Poisoning by thyroid and thyroid derivatives
962.8	Poisoning by antithyroid agents
E932.0	Adrenal cortical steroids causing adverse effect in therapeutic use
E932.4	Anterior pituitary hormones causing adverse effect in therapeutic use
E932.5	Posterior pituitary hormones causing adverse effect in therapeutic use
E932.6	Parathyroid and parathyroid derivatives causing adverse effect in therapeutic use
E932.7	Thyroid and thyroid derivatives causing adverse effect in therapeutic use
E932.8	Antithyroid agents causing adverse effect in therapeutic use

**Miscellaneous diagnoses that indicate growth problem during childhood**

V21.0	Period of rapid growth in childhood
V54.02	Encounter for lengthening/adjustment of growth rod
783.4	Lack of expected normal physiological development in childhood
783.40	Lack of normal physiological development, unspecified

**Diagnoses of genetic/chromosomal & congenital diseases that affect height—patient will be excluded if diagnosis is found anywhere in EMR**

253.3	Pituitary dwarfism
259.4	Dwarfism, not elsewhere classified
733.91	Arrest of bone development or growth
756.4	Chondrodystrophy
756.51	Osteogenesis imperfecta
758.0	Down's syndrome
758.6	Gonadal dysgenesis
759.81	Prader-Willi syndrome
759.82	Marfan's syndrome
243	Congenital hypothyroidism
759.1	Congenital anomalies of adrenal gland
759.89	Other specified congenital anomalies, so described
138	Post Poliomyelitis Syndrome

**Diagnoses in adulthood that may affect height or make measurements inaccurate--exclude height measurements that occur on or after these diagnoses****Lower limb amputation**

897	Traumatic amputation of leg(s) (complete) (partial)
897.0	Traumatic amputation of leg(s) (complete) (partial), unilateral, below knee, without mention of complication
897.1	Traumatic amputation of leg(s) (complete) (partial), unilateral, below knee, complicated
897.2	Traumatic amputation of leg(s) (complete) (partial), unilateral, at or above knee, without mention of complication
897.3	Traumatic amputation of leg(s) (complete) (partial), unilateral, at or above knee, complicated
897.4	Traumatic amputation of leg(s) (complete) (partial), unilateral, level not specified, without mention of complication
897.5	Traumatic amputation of leg(s) (complete) (partial), unilateral, level not specified, complicated
897.6	Traumatic amputation of leg(s) (complete) (partial), bilateral (any level), without mention of complication
897.7	Traumatic amputation of leg(s) (complete) (partial), bilateral (any level), complicated

V49.7	Lower limb amputation status
V49.70	Lower limb amputation, unspecified level
V49.73	Lower limb amputation, foot
V49.74	Lower limb amputation, ankle
V49.75	Lower limb amputation, below knee
V49.76	Lower limb amputation, above knee
V49.77	Lower limb amputation, hip

### Osteoporosis & other bone deforming disorders

733.0	Osteoporosis
733.00	Osteoporosis unspecified
733.01	Senile Osteoporosis
733.02	Idiopathic Osteoporosis
733.03	Disuse Osteoporosis
733.09	Other Osteoporosis
588.0	Renal osteodystrophy
722.4	CERVICAL DISC Degeneration
722.5	Degeneration of thoracic or lumbar intervertebral
722.51	THORACIC DISC Degeneration
722.52	LUMB/LUMBOSAC DISC Degeneration
722.6	Degeneration of intervertebral disc, site unspecified
737.10	Kyphosis (Acquired) (Postural)
737.30	Scoliosis (and Kyphoscoliosis), Idiopathic

**Comment [JSA1]:** Can cause lowered bone density like osteoporosisdoes

### Vertebral fracture

733.13	Pathologic fracture of vertebrae
805	Fracture of vertebral column without mention of spinal cord injury
805.0	Closed fracture of cervical vertebra without mention of spinal cord injury
805.00	Closed fracture of cervical vertebra, unspecified level without mention of spinal cord injury
805.01	Closed fracture of first cervical vertebra without mention of spinal cord injury
805.02	Closed fracture of second cervical vertebra without mention of spinal cord injury
805.03	Closed fracture of third cervical vertebra without mention of spinal cord injury
805.04	Closed fracture of fourth cervical vertebra without mention of spinal cord injury
805.05	Closed fracture of fifth cervical vertebra without mention of spinal cord injury
805.06	Closed fracture of sixth cervical vertebra without mention of spinal cord injury
805.07	Closed fracture of seventh cervical vertebra without mention of spinal cord injury
805.08	Closed fracture of multiple cervical vertebrae without mention of spinal cord injury
805.1	Open fracture of cervical vertebra without mention of spinal cord injury
805.10	Open fracture of cervical vertebra, unspecified level without mention of spinal cord injury
805.11	Open fracture of first cervical vertebra without mention of spinal cord injury
805.12	Open fracture of second cervical vertebra without mention of spinal cord injury
805.13	Open fracture of third cervical vertebra without mention of spinal cord injury
805.14	Open fracture of fourth cervical vertebra without mention of spinal cord injury

**Comment [JSA2]:** Instead of these fracture diagnoses, can optionally use NLP to determine if vertebral compression fractures occurred, as no specific ICD-9 code for this type of fracture

805.15	Open fracture of fifth cervical vertebra without mention of spinal cord injury
805.16	Open fracture of sixth cervical vertebra without mention of spinal cord injury
805.17	Open fracture of seventh cervical vertebra without mention of spinal cord injury
805.18	Open fracture of multiple cervical vertebrae without mention of spinal cord injury
805.2	Closed fracture of dorsal (thoracic) vertebra without mention of spinal cord injury
805.3	Open fracture of dorsal (thoracic) vertebra without mention of spinal cord injury
805.4	Closed fracture of lumbar vertebra without mention of spinal cord injury
805.5	Open fracture of lumbar vertebra without mention of spinal cord injury
805.8	Closed fracture of unspecified part of vertebral column without mention of spinal cord injury
805.9	Open fracture of unspecified part of vertebral column without mention of spinal cord injury
806	Fracture of vertebral column with spinal cord injury
806.0	Closed fracture of cervical vertebra with spinal cord injury
806.1	Open fracture of cervical vertebra with spinal cord injury
806.2	Closed fracture of dorsal (thoracic) vertebra with spinal cord injury
806.3	Open fracture of dorsal vertebra with spinal cord injury
806.8	Closed fracture of unspecified vertebra with spinal cord injury
806.9	Open fracture of unspecified vertebra with spinal cord injury
V54.17	Aftercare for healing traumatic fracture of vertebrae
V54.27	Aftercare for healing pathologic fracture of vertebrae
805.50	FRACTURE OF VERTEBRAL COLUMN W/O MENTION OF SPINAL CORD INJURY;LUMBAR,OPEN;CERVICAL VERTEBRA,UNSPECIFIED LEVEL
336.9	Unspecified disease of spinal cord

**Comment [JSA3]:** This ICD-9 code inc. spinal cord compression

**Miscellaneous diagnoses indicating height reduction in adulthood after attaining full height**

781.91	Loss of height
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