

# Ellis-Van Creveld Syndrome



By Gabriel Washington

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Chondroectodermal  
dysplasia



Six-fingered  
Dwarfism

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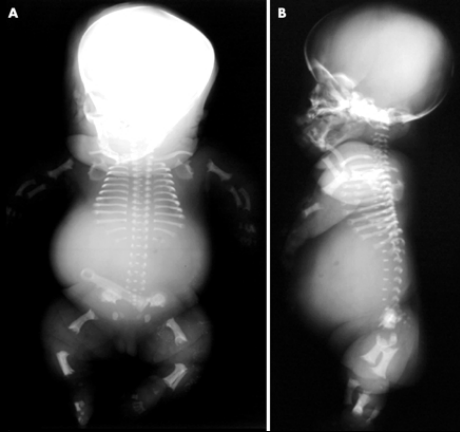


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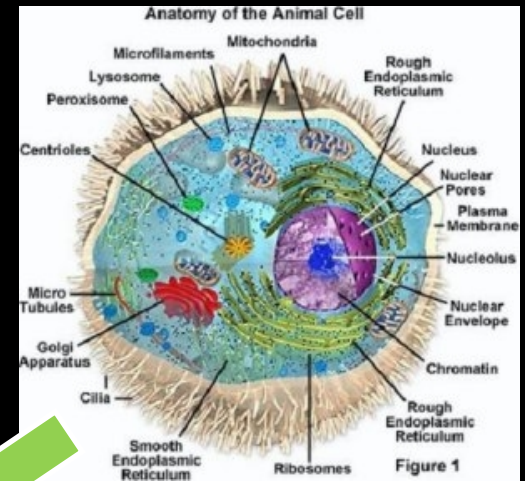
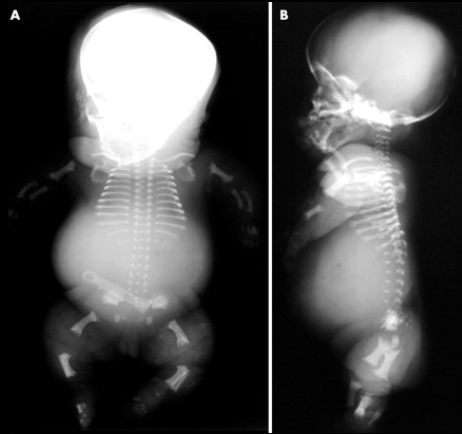
# Ellis-Van Creveld Syndrome



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# Signs of Ellis Van Creveld

## The New York Times

### Symptoms

- Cleft lip or palate
- Epispadias or undescended testicle (cryptorchidism)
- Extra fingers (polydactyly)
- Limited range of motion
- Nail problems, including missing or deformed nails
- Short arms and legs, especially forearm and lower leg
- Short height (between 3 1/2 and 5 feet tall)
- Sparse, absent, or fine textured hair
- Tooth abnormalities:
  - Peg teeth
  - Widely spaced teeth
  - Teeth present at birth (natal teeth)
  - Delayed or missing teeth



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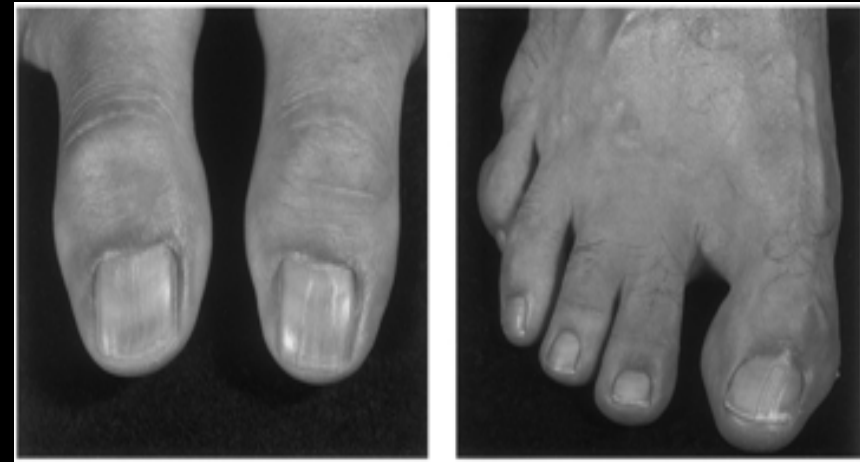
Fig. 1 : Short stature & genu valgum

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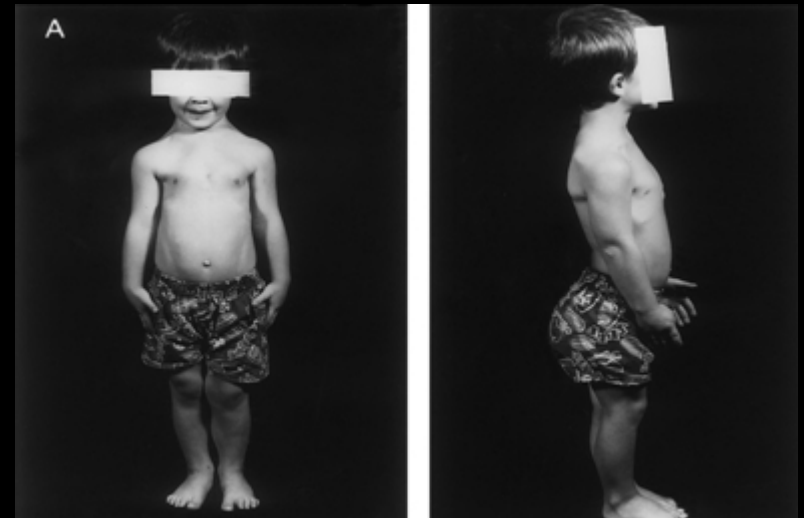


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# Diagnosis

## **Exams and Tests**

Signs of this condition include:

- Growth hormone deficiency
- Heart defects, such as a hole in the heart (atrial septal defect) occur in about half of all cases

Tests include:

- Chest x-ray
- Echocardiogram
- Genetic testing may be available for mutations in the EVC gene
- Skeletal x-ray
- Ultrasound
- Urinalysis

# Treatment

## Treatment List for Ellis-van Creveld syndrome

The list of treatments mentioned in various sources for [Ellis-van Creveld syndrome](#) includes the following list. Always seek professional medical advice about any treatment or change [in treatment](#)<sup>\*</sup> plans.

- Multidisciplinary management
- [Supportive care](#)
- Dietary counselling
- Dental care-oral hygiene instruction, plaque control
- Prosthetic rehabilitation
- Orthopedic procedures correct polydactyly and other orthopedic malformations
- Cardiac surgery may be needed to correct cardiac anomalies
- Thoracic expansion has been attempted in some patients
- Urologic surgery is required if epispadias, cryptorchidism, or both are present

# Prevention

Genetic counseling can aid couples in making informed decisions about pregnancies





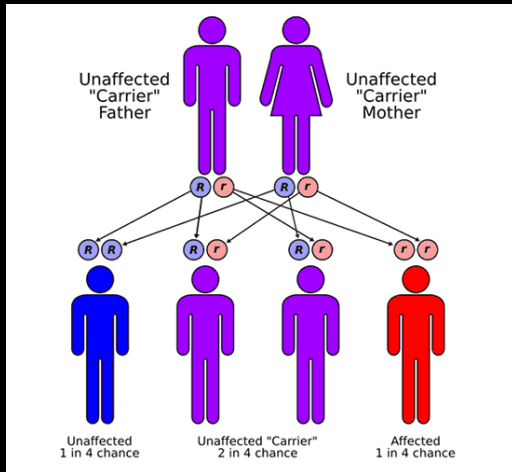
# Where can it be found?



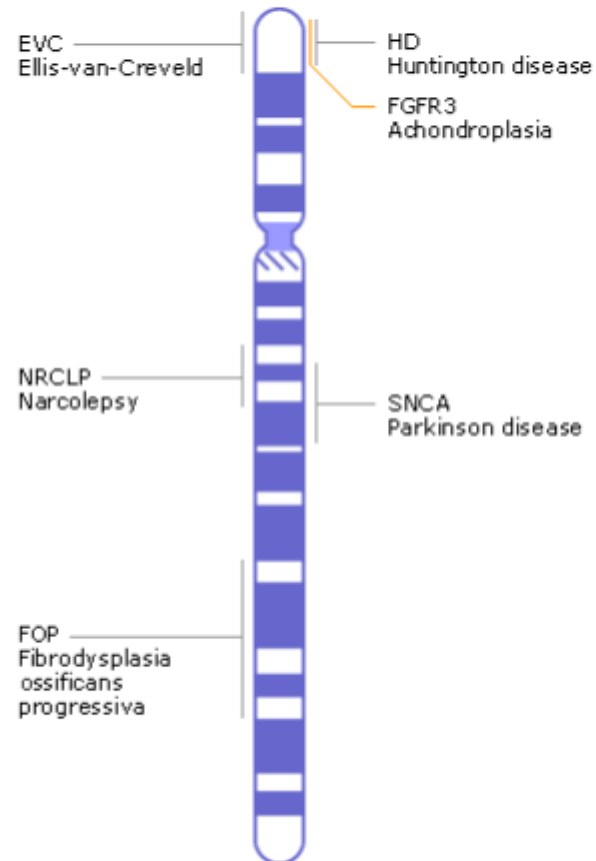
Lancaster County,  
Pennsylvania

Old Order Amish Population

# ... more specifically



- Contains approximately 1600 genes
- Contains approximately 190 million base pairs, of which ~95% have been determined
- See the diseases associated with chromosome 4 in the MapViewer



5,700 K    5,710 K    5,720 K    5,730 K    5,740 K    5,750 K    5,760 K    5,770 K    5,780 K    5,790 K    5,800 K    5,810 K    5,820 K    5,830

assembly - Sequence NC\_000004.11: Homo sapiens chromosome 4, GRCh37.p2 prim

- SNP

- Cited Variants

- NHGRI GWAS Variants

- NCBI genes

1    2 1 2    1    1    2    2

rs34624556    rs667338.3    rs6820907

EVC2

NM\_001166136.1

NP\_001159608.1

4520

NM\_147127.4

rs34624556

NP\_667338.3

4520    rs6820907

LOC100507314

XR\_108541.1

EVC

NM\_153717.2

NP\_714928.1

rs2291157    rs16837598    rs6414624    rs112608085    rs41269557

- NG Alignments

NG\_008843.1

Genomic context 🔍 ?

chromosome: 4; Location: 4p16 [See EVC in MapViewer](#)

# What's the problem?

Mutation in the EVC or EVC2 gene

Premature termination codon



# Citations

About.com “Ellis-van Creveld”

<http://rarediseases.about.com/cs/ellisvancreveld/a/111603.htm>

“Founder Effect” definition

[http://en.wikipedia.org/wiki/Founder\\_effect](http://en.wikipedia.org/wiki/Founder_effect)

Healthline “skeletal dysplasia”

<http://www.healthline.com/galecontent/skeletal-dysplasia>

KidsHealth “cleft lip”

[http://kidshealth.org/parent/medical/ears/cleft\\_lip\\_palate.html](http://kidshealth.org/parent/medical/ears/cleft_lip_palate.html)

NCBI “EVC”

<http://www.ncbi.nlm.nih.gov/books/NBK22264/>

NCBI Prenatal Diagnosis

<http://www.ncbi.nlm.nih.gov/pubmed/9621386>

NCBI Chromosome picture

<http://www.ncbi.nlm.nih.gov/books/bookres.fcgi/gnd/ch17d1.pdf>

New York Times Health Guide

<http://health.nytimes.com/health/guides/disease/ellis-van-creveld-syndrome/overview.html>

OMIM “Ellis-van Creveld

<http://www.ncbi.nlm.nih.gov/omim/225500>

WebMD “treatment and diagnosis”

[http://www.wrongdiagnosis.com/e/ellis\\_van\\_creveld\\_syndrome/treatments.htm](http://www.wrongdiagnosis.com/e/ellis_van_creveld_syndrome/treatments.htm)