

## Dwarfism in Miniature Horses

- A. Phenotypical Characterization
- B. Compared to Human types
- C. Phenotype vs. Genotype
- D. Old and New sub-types

### Current hypotheses / theories on mutant gene transmission

- All dwarf types in Miniatures are currently thought to be passed by autosomal recessive transmission – non-sex linked and only expressed in homozygous recessive form.
- There is some thought to possible co-dominance in certain types due to gene interaction showing expressivity in some individuals

### Inheritance of mutant gene

- Mutations are autosomal, both sexes can be dwarfs. If mutations are recessive as thought; both parents pass on one copy each to the foal and therefore the foal is a homozygous recessive receiving two copies.
- Current population statistical analysis shows inheritance as an autosomal recessive mutation

## Dwarf Phenotypes compared to human

- Possible homologous human types seen in miniature horses or more?

### Type 1

- Achondroplasia, Hypochondroplasia, pseudoachondroplasia, Acromesomelic Dysplasia

### Type 2

- Femoral Hypoplasia, pseudoachondroplasia

### Type 3

- Kniest Syndrome, Dyggve-Melchior-Clausen Syndrome, Diastrophic Dwarfism

### Type 4

- Achondrogenesis, Hypochondrogenesis, Type IA, IB
  - Diastrophic Dysplasia

## Achondroplasia characteristics

- The limbs have rhizomelic shortening
- The facial features include a large head with a prominent forehead. The midface is often small with a flat nasal bridge and narrow nasal passages
- Disproportionate short stature
- Airway obstruction can be "central" in origin (due to foramen magnum compression) or "obstructive" in origin (due to narrowed nasal passages). Symptoms of airway obstruction include snoring

## Acromesomelic Dysplasia

- Craniofacial- Disproportionately large head with relative frontal prominence, with or without relatively short nose
- Limbs- short limbs with short hands and feet, bowed forearms that are relatively shorter than upper arms, limited elbow extension
- Spine – development of lower thoracic kyphosis
- **Autosomal recessive**

### Pseudoachondroplasia

- Craniofacial – normal head size and face
- Limbs- Disproportionately short.
- Growth – postnatal onset of short-limbed growth deficiency that becomes obvious between 18 mos – 2 yrs.

### Femoral Hypoplasia

- Growth – small stature, predominantly the result of short limbs
- Facial – some normal, some unusual with short nose, micrognathia, other deformities
- Limbs bilateral, usually asymmetrical involvement, Hypoplastic to absent femora and variable asymmetric involvement of fibula and tibia. Variable hypoplasia of humeri with restricted elbow movement, including radioulnar and radiohumeral synostosis
- Pelvis – hypoplastic acetabulae, constricted iliac base with vertical ischial axis, large obturator foramina
- Spine – scoliosis, dysplastic sacrum

### Kniest Syndrome

- Growth – disproportionate short stature with short barreled shaped chest
- Craniofacial – flat faces with prominent eyes, low nasal bridge, myopia with retinal detachment and degeneration, head which is of normal size, is relatively large.
- Limbs – enlarged joints with limited joint mobility and variable pain and stiffness, short limbs with bowing, some irregularity of epiphyses with late ossification of femoral heads. Flexion contractures of hips, small pelvis.
- Defect in COL2A1 different mutation of gene than type 4

Miniature Type 4 Achondrogenesis, Hypochondrogenesis, Type IA, IB  
- Diastrophic Dysplasia

- Growth – extremely small stature
- Craniofacial – Cranium large for gestational age, low nasal bridge, micrognathia
- Limbs – severe micromelia
- Bones poorly ossified for gestational stage, ribs extremely short and thin
- Lethal recessive
- Mutation is in gene for diastrophic dysplasia which codes for a sulfate transporter DTDST

Miniature Type 4 Achondrogenesis, Hypochondrogenesis, Type IA, IB  
- Diastrophic Dysplasia



Chondrodysplastic dwarfism in Japanese brown cattle is an autosomal recessive disorder characterized by short limbs. Previously, it was mapped to the locus responsible for the disease on the distal end of bovine chromosome 6. Here the region is approximately 2 cM by using linkage analysis, constructed a BAC and YAC contig covering this region, and identified a gene, LIMBIN (LBN), that possessed disease-specific mutations in the affected calves. One mutation was a single nucleotide substitution leading to an activation of a cryptic splicing donor site and the other was a one-base deletion resulting in a frameshift mutation. Strong expression of the Lbn gene was observed in limb buds of developing mouse embryos and in proliferating chondrocytes and bone-forming osteoblasts in long bones. These findings indicate that LBN is responsible for bovine chondrodysplastic dwarfism and has a critical role in a skeletal development

Congenital lethal chondrodysplasia was studied in two female Dexter fetuses aborted mid to late gestation. Clinicopathological findings including histological changes in limb bones, and analysis of pedigree information were evaluated.

**RESULTS:** Characteristic features of congenital lethal chondrodysplasia (Dexter bulldog) include abortion, disproportionate dwarfism, a short vertebral column, marked micromelia, a relatively large head with retruded muzzle, cleft palate and protruding tongue and a large abdominal hernia. Histological changes in limb bones are consistent with failure of endochondral ossification. Dexter chondrodysplasia is considered to be inherited in an incompletely dominant manner with the homozygous form producing the congenital lethal condition.

**If you want to send samples or donate to my research through UK**

1. From live dwarf miniature

Send to Attn: John Eberth/ Ernest Bailey –

Dept of Veterinary Science 108 Gluck Equine Research Center University of Kentucky  
Lexington KY 40546-0099

- A. Picture of animal (very important so I can determine types of dwarfs)
- B. Pictures of parents too is great
- C. 2 tubes of blood in lavender or yellow topped tubes. Label tubes with "dwarf"
- D. 2 tubes of blood as above from sire and dam if possible. Label "sire" and "Dam"
- E. Pedigree of sire and dam if you are willing to send it.
- F. Ship UPS 2nd day

If you want to make a donation and to be tax deductible you can make it out to;

- 1. AMHA for Genetic Research on Miniature Horse dwarf defects or
- 2. University of Kentucky at Lexington for Genetic Research on Miniature Horse dwarf defects.

**If you want to donate Carcasses : Normal or Dwarf**

Please contact me directly for specific  
freezing and transporting instructions.

John E. Eberth  
Arion Management Inc.  
970 Hifner Rd. Versailles, KY 40383  
arionmt@aol.com  
859-879-6344  
859-948-8106

**If you want to send samples or donate to my research through TAMU**

- Dr Cothran at Texas A&M After April
- The checks need to be made payable to "TAMU Foundation" they need to be mailed to

Merrie Noak  
Business coordinator I  
Department of Veterinary Integrated Biosciences  
Texas A&M University  
4458 TAMU  
College Station TX 77843-4458  
Attn: Jerry Zalmanek

The accompanying letter should state that the funds are to earmarked for the benefit of research endeavors in the area of Miniature horse dwarfism and there are no stipulations on their use. Once an account number has been established it would be helpful to have it listed in the letter as well.

## Information to Send Donations Only

- If you want make a donation that is tax deductible you can make the checks payable to :
- 1. AMHA for genetic research on Miniature horse dwarf defects and there are no stipulations on their use. These donations will be put into a Genetic Research Fund. OR
- 2. University of Kentucky at Lexington for genetic research on Miniature horse dwarf defects and there are no stipulations on their use. OR
- 3. TAMU the accompanying letter should state that the funds are to earmarked for the benefit of research endeavors in the area of Miniature horse dwarfism and there are no stipulations on their use. Once an account number has been established it would be helpful to have it listed in the letter as well.

## Thank You

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