Differential diagnosis of pre- and postnatal short stature revisited: 3-M Syndrome

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INTRODUCTION

- synonyms: 3-M Syndrome, Three-M Slender Boned Nanism, Yakut Short Stature Syndrome
- rare autosomal recessive disorder, first described by Miller, McKusick and Malvaux
- frequency < 1/1,000,000, prevalence 200 reported cases

CRITERIA

- Postnatal: U2 shortened long bones, 
- Pre- and postnatal disproportionate short stature, Micromelia, relative hypoplasia of the thorax, 
- Facial anomalies, characteristic dysmorphism

RESULTS

Findings:

- Birth weight: 3.1 kg (<3.P, [70th])
- Birth height: 44 cm (<3.P, [70th])
- Head circumference: 33.5 cm (10th)
- Body length: 44 cm (<3.P, [70th])
- Ratio of subischial leg length to true height 0.76 (<3 SDS, normally increased)

Phenotypic findings:

- Facial anomalies:
  - Micrognathia, pointed chin
  - Blepharophimosis
  - Absent or hypoplastic labial frenum
- Head:
  - Long (67 cm)
  - Hypotelorism
  - DOWN syndrome
  - Augmented head circumference
- Trunk:
  - Upright posture
  - Micromelic hands
- Abnormalities:
  - Hypoplastic clavicles
  - Hypoplastic ribs
- Postnatal:
  - U2 shortened long bones

TREATMENT:

- Improvement of short stature through surgical bone lengthening or growth hormone substitution therapy
- skeletal abnormalities: orthopedic techniques and surgery
- Dental anomalies: orthodontic procedures

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CONCLUSIONS

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Children with 3M syndrome develop disproportionate short stature with conspicuous radiological signs. The diagnosis is confirmed by detection of mutations in one of the three genes: CCDC8, OBSL1 or CUL7.

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CASE REPORT

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