Dysphagia and Language Impairment in King-Denborough Syndrome

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Background

Aged 3;0 Haider was referred to speech and language therapy by the Paediatrician. Assessment showed that he was only using single word nouns in English and two-word combinations (verb + patient) in Mirpuri. Audiology reported hearing within normal limits and the Educational Psychology Service reported average non-verbal abilities. A diagnosis of language impairment (LI) was made and Haider was offered a place at the local language unit. At this time it was recognised that he had subtle gross motor difficulties and deformity of the spine and sternum. Haider was discharged from the speech and language therapy service aged 7;1 as his communication skills were satisfactory. King-Denborough syndrome was not diagnosed until age 10;0 following a malignant hyperthermia episode during surgery: a reaction to anaesthesia which is a recognised feature of this syndrome.

Aged 3;4 Bilal was referred to speech and language therapy by the Community Paediatrician. At this time the speech and language therapist discussed the possibility that Bilal had the same syndrome as his older brother. This was confirmed following genetic tests. Bilal was able to follow simple instructions, had no recognisable words and some vocalisations. A diagnosis of LI was made and Bilal attended the language unit. Bilal continues to require speech and language therapy.

Language Impairment

Both children presented with severe and complex language impairment.

- Haider attended the local language unit for Year 2.
- Bilal attended the language unit for Years 1 and 2.

Mother Tongue Therapy

Both children were exposed to Mirpuri, a Pakistani heritage language at home. In line with professional guidelines and current evidence based practice, both children received direct therapy in mother tongue only. The children's English acquisition was allowed to develop by exposure to the classroom environment. On discharge from the language unit both children had acquired English skills which matched their mother tongue skills.

www.speechtherapy.co.uk and www.bilingualism.co.uk

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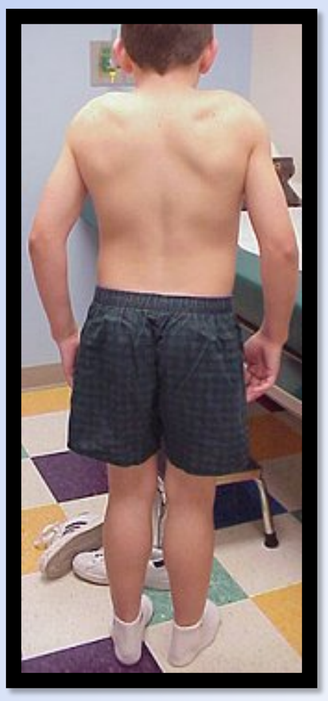
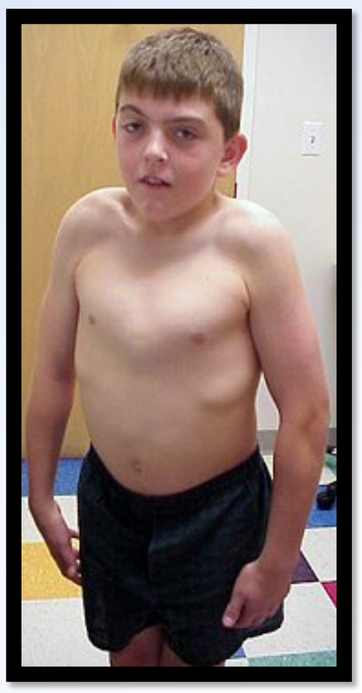
Features

(King *et al.* 1972, Graham *et al.* 1998)

The syndrome is an autosomal dominant or sporadic genetic disorder characterised by:

- Malignant hyperthermia (triggered by excess heat or anaesthetic)
- Weakness (including delayed motor development and scapular winging)
- Phenotypical features (physical features) including:
- Skeletal abnormalities (short stature and pectus carinatum/excavatum)
- Facial dysmorphia
- **NOT previously reported:**
- Severe language impairment
- Dysphagia





Images from http://neuromuscular.wustl.edu/msys/myoglob.html#kd

Dysphagia

Haider was re-referred by the Paediatrician aged 13;0 with dysphagia.

Haider reported a one year history of painful swallowing and nausea. He had a recent chest infection, was coughing at night and had lost weight. A videofluoroscopy showed fluids were spilling into the pharyngeal area. Haider was using a chin-tuck to clear the bolus from the oral cavity. Repeated swallows indicated fatigue leading to an uncoordinated swallow. A gastrostomy was discussed but rejected by Haider. Currently Haider remains under the care of the multidisciplinary team. He has daily nutritional supplements and his weight and height remain at the 2nd centile.

Bilal has not as yet presented with dysphagia.