When you are ready for more information, Unique may be able to help. We can answer individual queries and we also publish a more detailed leaflet about duplications of 7q. This leaflet is not a substitute for personal medical advice. Families should consult a medically qualified clinician in all matters relating to genetic diagnosis, management and health. The information is believed to be the best available at the time of publication and the content of the full leaflet on which this text was based was verified by Dr Steve Scherer, The Hospital for Sick Children, Ontario, Canada and by Professor Maj Hulten, Professor of Medical Genetics, University of Warwick, 2005.

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When a chromosome disorder is as rare as a 7q duplication, it is not possible to be certain how it will affect an individual child. Very few babies have been described with a duplication (a trisomy) of the entire long arm of chromosome 7. In babies and children with a smaller 7q duplication, the most common features are also common in children with other chromosome disorders.

- Developmental delay
- Some degree of learning difficulty or disability
- Low muscle tone, so the body feels floppy
- Low birth weight and slow weight gain in babies. Eventual height is often short, although not always for those with a small duplication at the end of the chromosome
- High palate (roof of the mouth). There may be a split (cleft) in the soft or hard part of the palate
- Noticeably large head and, at birth, a very large soft spot (fontanelle) on top
- Most babies are healthy at birth, although there may be a heart condition. Occasionally the structure of the brain is affected
- Unusual position of one or both feet (talipes, club foot)
- Curvature of the spine
- Unusual facial features. Most of these are of no consequence to the child and are usually quite subtle. They might include a high, rounded forehead, downslanting and wide set eyes, skinfolds across the inner corners, squint (strabismus) and low set ears; frequent respiratory infections in children.

Duplications from 7q31 to the end of the chromosome

The rate of health problems in the first year of life seems to be high. Common features, most of them frequent in children with other chromosome disorders, include: low birth weight; developmental delay; learning difficulties; growth delay; feeding difficulties; unusual brain structure; heart problems; underdeveloped lungs; cleft palate; unusually large fontanelle (soft spot); skeletal anomalies, for example, missing 12th ribs.

Duplications from 7q32-7q35 to the end of the chromosome

Babies and children with this duplication, often linked with a deletion from another chromosome as part of an unbalanced chromosome translocation, appear to do better than those with a larger duplication extending into band 7q31. The most consistent features are quite non-specific and include a birth weight within the normal range and subsequent normal growth rate, a variable degree of developmental delay and learning difficulty, a relatively large head, some feeding difficulties and some unusual genital features in boys such as undescended testicles.

Duplications from 7q36 to the end of the chromosome

There is little experience with individuals with a small duplication of material near the end of the chromosome, but it appears that effects are less far-reaching than when the duplication is larger. Common features include:

- Some degree of developmental delay, especially speech
- Large head with a prominent forehead.