# Large Deletion of 5q12 With Dysmorphic Features, Poor Growth, Delays, Behavioral Anomalies, and Seizures: A Case Report and Literature Review

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## Background

5q12 deletion syndrome (OMIM #615668) is a rare condition with only thirteen individuals that have been reported in the literature. Though the DECIPHER database includes fifty entries of deletions in this region, our patient's rare deletion remains larger than all but 3 entries in the database, most of which are less than 1 Mb and are of benign or uncertain significance. Describing our patient's clinical features and comparing them to those in the literature will help to further elucidate on 5q12 deletion syndrome and the clinical consequences of such a rare deletion.

| | Our Patient | Literature (13 cases) |
|---|---|---|
| Sex | F | 8F and 5M |
| Pregnancy complications | - | 3/13 |
| Post-natal growth restriction | + | 6/13 |
| Seizures | + | 6/13 |
| Muscle tone anomalies | - | 5/13 |
| Intellectual disability | + | 7/13 |
| Developmental delays | + | 9/13 |
| Behavioral anomalies | ADHD, anxiety | 4/13 |
| Thin habitus | + | 3/13 |
| Palpebral fissure anomalies | up-slanting (almond-shaped eyes) | 4/13 |
| Ptosis | - | 3/13 |
| Esotropia | - | 3/13 |
| Visual impairment | + | 3/13 |
| Astigmatism | - | 2/13 |
| Hypermetropia | - | 2/13 |
| Myopia | + | 1/13 |
| Heterochromia | partial, OD | - |
| Prominent columella | - | 3/13 |
| Large forehead | - | 4/13 |
| Philtrum anomalies | - | 4/13 |
| Coarse facies | - | 4/13 |
| Flat facies | - | 3/13 |
| Micrognathia | + | 3/13 |
| Nasal anomalies | - | 8/13 |
| Thin upper lip | - | 3/13 |
| Ear anomalies | - | 5/13 |
| Limb anomalies | - | 6/13 |
| Short neck | - | 2/13 |
| Frontal prominence/bossing | - | 2/13 |
| Microcephaly | + | - |
| Large teeth with malocclusion | + | 1/13 |
| Hair anomalies | - | 2/13 |
| Recurrent infections | - | 3/13 |
| Cardiovascular abnormalities | - | 2/13 |
| Low BP | - | 2/13 |
| Asthma | + | - |
| Other (1 each) | - | brachycephaly, eyelid angioma, hypertelorism, hypotelorism, epicanthus, strabismus, exotropia, deep-set eyes, large mouth, macroglossia, ovarian cyst, cryptorchidism, scoliosis, sacral dimple, congenital hip dislocation, brachydactyly, hexadactyly, flat feet, ataxia, atypical nevi, delayed puberty, anemia |

Table 1: Clinical features of our patient as compared to the thirteen patients currently in the literature with pathogenic deletions.

## Case Presentation

The patient was born at 42 weeks via spontaneous vaginal delivery to a non-consanguineous 34-year-old G3P1011>2 mother and 40-year-old father. One ultrasound was reported to reveal slow growth, but further ultrasounds revealed no anomalies. There were no prenatal exposures and no neonatal complications. At 10 years old, she has delays and intellectual disability with behavioral problems and seizures. Her dysmorphic features include almond-shaped eyes with up-slanting palpebral fissures, right iris heterochromia, large teeth with malocclusion, microcephaly, and micrognathia with short stature and thin habitus. Her family history is significant for ADD in her maternal uncle and in her mother, and allergic rhinitis in her mother, father, and sister. Chromosomal SNP microarray revealed a pathogenic 13.7Mb deletion of 5q12 to q13.3, overlapping the region of 5q12 deletion syndrome.

## Methods

Deletions were identified by clinical SNP microarray and analyzed according to the GRCh37 genome build.

## Results

A pathogenic 13,652 kb deletion of the long arm of chromosome 5 involving bands 5q12.1 to 5q13.2 was detected. This deletion is consistent with the rare chromosome 5q12 deletion syndrome (OMIM #615668) which has been described in thirteen patients. The reported cases all have inconsistent breakpoints and varying deletion sizes from 0.9Mb to as large as 17.2 Mb. The clinical features of 5q12 deletion syndrome are also varied, but commonly include growth restriction, intellectual disability, behavioral abnormalities, non-specific ocular defects (including esotropia, strabismus, and ptosis), micrognathia, prominent columella, long palpebral fissures, hypotonia, delayed speech, autistic features, decreased body mass, low blood pressure, and long fingers and toes. Seizures have also been reported in 5q12 deletion syndrome.

## Conclusions and Discussion

Our patient's clinical picture involves post-natal growth restriction, seizures, intellectual disability, developmental delays, behavioral anomalies, up-slanting palpebral fissures, right iris heterochromia, visual impairment, myopia, micrognathia, microcephaly, thin habitus, large teeth with malocclusion, and asthma. Behavioral anomalies, growth restriction, intellectual disability, developmental delay, seizures, ocular defects, and certain dysmorphic features correspond to both our patient and those in the literature. Findings in our patient that are not seen in current literature include up-slanting palpebral fissures, right iris heterochromia, microcephaly, and asthma. Outlining our patient's clinical findings compared to those of the only thirteen other individuals noted in the literature sheds needed light on the clinical significance of such a rare deletion and its host of potential features.

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