

## 3p Deletion Syndrome

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3p deletion is a rare cytogenetic finding. Here we describe a 3 months old male with congenital malformations. His karyotype revealed 3p deletion 46,XY,del(3)(p25-pter). The child had flexion deformity of wrist and elbow which has never been reported before.

**Keywords:** 3p deletion, trigonocephaly, micrognathia.

The 3p deletion syndrome has been proposed as a contiguous gene syndrome with the spectrum of defects depending upon the overall size of the deleted segment [1]. The phenotype of individuals with 3p deletions varies from normal to severe. The 3p25 chromosome deletion syndrome was first reported in 1978 [2]. Since the first case, less than 50 cases with distal 3p deletions have been reported. Characteristic features of the syndrome include low birth weight, microcephaly, trigonocephaly, hypotonia, mental and growth retardation, ptosis and micrognathia. Other features that may be seen include polydactyly, renal anomalies, congenital heart defects, ear anomalies, and gastrointestinal tract anomalies. It has been suggested that a 1.5 Mb minimal terminal deletion including the two genes *CRBN* and *CNTN4* in chromosome 3 are sufficient to cause the syndrome. In addition the *CHLI* gene, mapping at 3p26.3 distally to *CRBN* and *CNTN4*, was proposed as candidate gene for a non specific mental retardation because of its high level of expression in the brain [3].

### CASE REPORT

A 3-months-old male child with congenital malformations was referred to us for cytogenetic investigations. Detailed pedigree analysis and in-depth evaluation of the clinical reports was undertaken. Chromosome preparations were made from peripheral lymphocytes using RPMI 1640 medium and phytohemagglutinin using standard method with modifications [4] and G-banding was done. Fifty metaphases were examined for numerical as well as structural abnormalities and five metaphases were karyotyped with Applied Imaging Software (Cytovision). A written consent was obtained from the parents before all the investigations.

A full term male child, weighing 2.10 kg born by caesarian section was presented with multiple congenital anomalies. The proband was the first child of healthy,

non-consanguineous parents. The mother had an uneventful pregnancy. He had a delayed and weak cry. At the time of examination, the infant was 3 months old with triangular face, hypertrichosis, bilateral exophthalmous eyeballs, depressed nasal bridge with wide nostrils, retrognathia and high arched palate. He presented bilateral flexion deformity of wrist and elbow, and calcaneovalgus deformity of right foot. He was lethargic, his neurological and motor milestones were delayed. He had history of neonatal asphyxia, cyanotic spells, recurrent vomiting and sleep apnea. X-ray skull showed partial closure of sutures. The CT scan revealed anteriorly pointed frontal closure of the metopic suture suggesting craniosynostosis with trigonocephaly. The venous sinuses were prominent and hyperdense. Routine peripheral blood film showed normochromic picture. Karyotyping of the case showed terminal deletion of the chromosome 3. The child expired a week after presentation.

### DISCUSSION

3p deletion syndrome is a rare disorder involving the short arm of chromosome 3. The clinical findings of our case were very severe and similar to the description in literature. The flexion deformity has previously not been reported in the literature. Karyotyping of the present case showed 46, XY, del(3)(p25-p26.34). Parents were not available for further investigations. This syndrome presents a strong connection between the severity of the disease and the portion of the deletion. Despite investigations of several genes in the 3p region involved in CNS development, a causative relationship between any particular transcript and the range of observed clinical manifestations has remained elusive. The minimal candidate region for 3p deletion, implicates haploinsufficiency of various genes and demonstrates the utility of high-resolution investigations of rare chromosomal rearrangements [6]. Some of the known genes in the 3p" phenotype have been previously described [1,5-10].

Karyotyping remains the gold standard for detecting chromosomal aberrations in cases with congenital anomalies. A meaningful correlation between the deletion and the clinical phenotype is not possible until further use of high-resolution investigations like CGH array to fully characterize the case, which was not possible due to financial constraints.

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## REFERENCES

1. Cargile CB, Goh DL, Goodman BK, Chen XN, Korenberg JR, Semenza GL, *et al.* Molecular cytogenetic characterization of a subtle interstitial del(3)(p25.3p26.2) in a patient with deletion 3p syndrome. *Am J Med Genet.* 2002;109:133-8.
2. Verjaal M, De Nef J. A patient with a partial deletion of the short arm of chromosome 3: karyotype: 46, XY, del(3)(p25). *Am J Dis Child.* 1978;132:43-5.
3. Cuoco C, Ronchetto P, Gimelli S, Béna F, Divizia MT, Lerone M, *et al.* Microarray based analysis of an inherited terminal 3p26.3 deletion, containing only the CHL1 gene, from a normal father to his two affected children. *Orphanet J Rare Diseases.* 2011;6:12.
4. Kaur A, Mahajan S, Singh JR. Cytogenetic profile of individuals with mental retardation. *Int J Hum Genet.* 2003;3:13-6.
5. Shuib S, McMullan D, Rattenberry E, Barber RM, Rahman F, Zatyka M, *et al.* Microarray based analysis of 3p25-p26 deletions (3p<sup>+</sup> syndrome), *Am J Med Genet. Part A.* 2009;149:2099-105.
6. Fernandez T, Morgan T, Davis N, Klin A, Morris A, Farhi A, *et al.* Disruption of *Contactin 4 (CNTN4)* results in developmental delay and other features of 3p deletion syndrome state *Am J Hum Genet.* 2004;74:1286-93.
7. Malmgren H, Sahlen S, Wide K, Lundvall M, Blennow E. Distal 3p deletion syndrome: detailed molecular cytogenetic and clinical characterization of three small distal deletions and review. *Am J Med Genet.* 2007;143A:2143-9.
8. Gunnarsson C, Foyn BC. Molecular characterization and clinical features of a patient with an interstitial deletion of 3p25.3-p26.1. *Am J Med Genet.* 2010;152 A:3110-4.
9. McCullough BJ, Adams JC, Shilling DJ, Feeney MP, Sie KCY, Tempel BL. 3p<sup>+</sup> syndrome defines a hearing loss locus in 3p25.3. *Hearing Research.* 2007;224:1-2:51-60.
10. Carayol J, Sacco R, Tores F, Rousseau F, Lewin P, Hager J, *et al.* Converging evidence for an association of ATP2B2 allelic variants with autism in male subjects. *Biological Psychiatry.* 2011; 170:880-7.

## Thoracoscopic Ligation of Thoracic Duct for Spontaneous Chylothorax

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Spontaneous chylothorax, without a predisposing factor is an uncommon cause of pleural effusion beyond the neonatal period. We present a case of left sided spontaneous chylothorax in a 20-month-old boy. We report successful management of this difficult problem with thoracoscopic ligation of thoracic duct after a failed trial with conservative management.

**Keywords:** *Chylothorax, Thoracoscopy, Thoracic duct, VATS.*

Chylothorax has various causes, including malignancy, trauma (including surgery), and miscellaneous disorders (such as deep vein thrombosis, sarcoidosis, and congestive heart failure), and can also be idiopathic [1,2]. Undetected malformations of the lymphatic trunks are implicated as a cause of spontaneous chyle accumulation. Management of spontaneous chyle accumulation in a child is a challenging task. We present a child with left sided spontaneous chylothorax who was managed with thoracoscopic ligation of thoracic duct on right side.

### CASE REPORT

A 20-month-old boy presented with fever and breathing difficulty for one week. There was no history of trauma or operative intervention in the child. The mother gave no history of excessive cough or vomiting. The child was otherwise healthy, with no significant past medical history. There was no history of recent trauma or history suggestive of cardiopulmonary disease. The child's immunization was up to date.

On examination, the child weighed 12 kg, was febrile,