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Clinical Images

Cri-du-chat syndrome

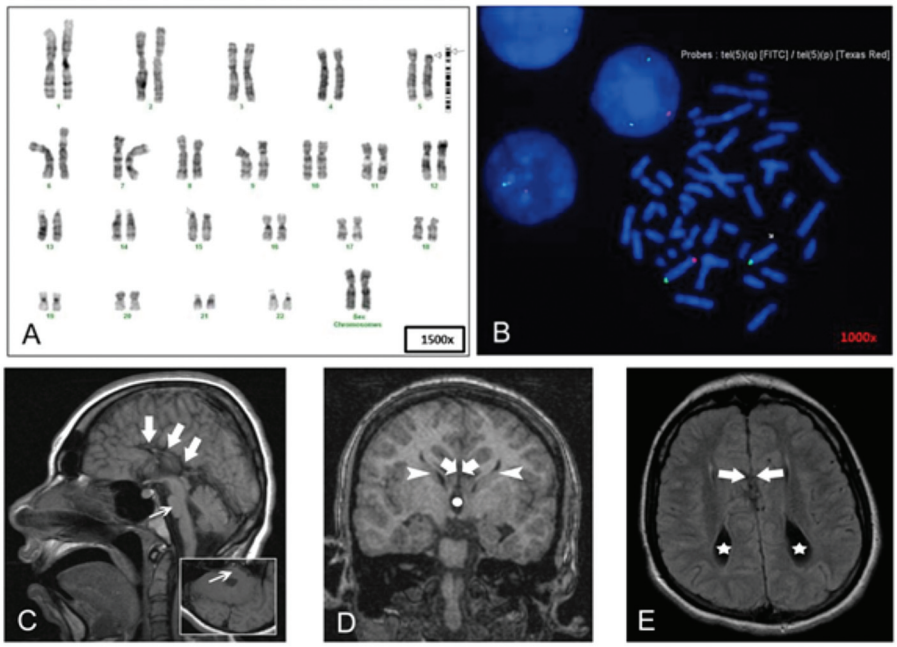

Fig. (A). G-banded karyotype (x 1500 magnification) showing 46,XX,del(5)(p15). Arrow indicates the breakpoint on p-arm of chromosome 5. **(B)**. FISH (x 1000 magnification) using telomeric probes (Aquarius) for chromosome 5p (red) and 5q(green). Arrow indicates the absence of FISH signal on 5p confirming the deletion. **(C-E)**. Brain MRI demonstrating complete corpus agenesis of corpus callosum (thick straight arrows in **C, D** and **E**), radial arrangement of medial hemispheric sulci, absence of cingulate sulcus, splaying of anterior part of the lateral ventricles (arrowheads in **D**), colpocephaly (stars pointing to the enlarged occipital horns of the lateral ventricle in **E**) and high riding third ventricle (a single circle in **D**) and pontine hypoplasia (a single thin straight arrow in **C** and inset).

A 19 year old girl, who presented to the Neurology unit of the department of Medicine, Sultan Qaboos University hospital (Oman) in July 2011, was evaluated for developmental delay, dysmorphic long narrow face, epicanthic folds, small ears, high pitched cat-like cry in early childhood and absence of expressive speech. Her metabolic work up for inborn error of metabolism was negative. Her karyotype (Fig. A) demonstrated 46,XX,del(5)(p15). Fluorescence *in situ* hybridization (Fig. B) showed telomeric deletion of 5p at region 15, confirming the diagnosis of Cri-du-chat syndrome[1,2]. Brain MRI (Figs. C-E) revealed complete agenesis of corpus callosum and pontine hypoplasia.

The most frequent imaging finding in Cri-du-chat syndrome, a rare chromosomal disorder resulting from 5p deletion, is pontine hypoplasia[1,2]. Our patient had the association of callosal agenesis that has been only rarely reported in the literature[3].

There is no specific therapy available for this condition[1,2]. The patient received symptomatic treatment *viz*. risperidone for her behaviour problems (with occasional violence directed against her relatives that was related to intellectual impairment) with some improvement and was referred for neuro-rehabilitation. She continued to have language problems at two years of follow up.

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