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Universitäts-Kinderkliniken · Eleonorenstiftung

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Zuerich, September 2, 2010/um

K O R K M A Z Sara Maria, 16.01.2010, Calea 13 Septembrie 107, Bl 103 Ap 22, 021689 Bukarest, Romania

This girl was seen as an out patient on August 30, 210, accompanied by the parents, following earlier email contact.

DIAGNOSIS: Joubert syndrome

History / actual situation

1st child, non related healthy parents. Details to the history have been kindly summarised in a letter by Dr. C. Iliescu. The diagnosis of Joubert syndrome (JS) has been confirmed by MRI, performed 14.05.2010. The mother has kindly sent me a copy before. This confirms undoubtedly the neuroimaging hallmarks of JS, namely vermis hypoplasia and molar tooth sign.

Over time the respiratory pattern has improved, less marked tachypnea. Normal respiration in sleep. No feeding difficulties. Reacts to sounds. Mild strabismus, but definitive visual contact. Sara is slow in her milestones, she has hypotonia, physiotherapy 3 times weekly was started at 2 ½ months.

Examination

Charming 7,5-month-old girl, weight 8,3 kg (P 50), length 68 cm (P 25), head circumference 43,7 cm (P 50). The facial appearance is well compatible with JS. Visual behaviour points to oculomotor apraxia. Definite visual contact and following are possible. Occasionally mild spontaneous horizontal nystagmus. Symmetrical spontaneous movements. Marked muscle hypotonia. No evidence of paralysis.

No additional abnormalities such as polydactyly, enoral frenula, tongue hamartoma.

Additional investigations

Abdominal ultrasound: Normal liver and kidneys.

helpSaraMaria.ro

Serum chemistry: Normal values for creatinine (37), AST (33), ALT (16). EDTA blood for DNA isolation was taken. This will be forwarded to the Lab of Prof. Valente in Roma, with whom I have a close cooperation.

Ophthalmological evaluation by Dr. V. Sturm (see separate letter)

Discussion

The following points were discussed:

- The diagnosis of JS (or Joubert syndrome and related disorders) is definitely confirmed.
- Up to now 10 genes are known, 9 autosomal, all accounting for less than 50 % of the patients. This also means that it is not sure that in reasonable time a mutation can be found in Sara.
- Prenatal diagnosis on bases of mutation analysis is only possible if a mutation is found in an index patient. Otherwise prenatal diagnosis depends on ultrasound / fetal MRI demonstrating an encephalocele and molar tooth sign, however, this is not reliably possible before week 20 to 22.
- Renal impairment is seen in a minority of patients, this tends to be a slow process. This means that testing of kidney function can be made at reasonably long intervals (say 2-3 years).

As Sara has clinically good visual function an ERG is not required.

It is not possible at this stage to give an individual prognosis for outcome. Developmental delay and subsequent evidence of congnitive impairment is seen in a great majority of patients, however, most patients learn to walk and truncal ataxia is hardly a problem on longterm observation, many patients get reasonable functional independence.

Best regards

Prof. Dr. med. E. Boltshauser, MD

- Ultrasount on CD
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Kopie:
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