Title

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Authors

Jonas, Roy E

Kimonis, Virginia E

Morales, Augusto

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Autosomal Recessive Syndrome Of Partial Agenesis Of The Corpus Callosum, Pontine Hypoplasia, White Matter Changes, Hypotonia, Mental Retardation, And Dysmorphic Features. † 349

Roy E Jonas, Virginia E Kimonis & Augusto Morales
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A brother and sister with dysmorphic features, severe developmental delays, and hypotonia who have associated partial agenesis of the corpus callosum, pontine hypoplasia, and progressive white matter abnormalities are described. Dysmorphic features include macrocrania, frontal bossing, deep set eyes, small palpebral fissures, and a curved nose. Discordant features include Duane's syndrome Type II, an ectopic right ureter, and strabismus in the male. Laboratory investigations included normal karyotype, serum amino acids, urine organic acids, carnitine, lactate, ammonia, leukocyte lysosomal enzyme studies, and CSF protein. Nerve conduction velocity and EEGs were normal. BAER revealed low amplitude brainstem waveforms suggestive of pontine abnormalities in the boy. The partial agenesis of the corpus callosum is atypical in that it affects not only the posterior splenium, but also the genu. This is clearly not explained by a late insult during development, as is usually suspected in partial agenesis. The pontine hypoplasia is also unique in that it has not been previously reported with an anatomically normal cerebellum. The white matter changes were located in the subcortex of the frontal lobes in MRI taken at about age 3, but they were not present during infancy. We found no evidence to support a previously specified leukodystrophy. The children are severely delayed developmentally, and we predict a regressive course. FG syndrome, Acrocallosal syndrome, and Andermann's syndrome, as well as case reports by Ciotti and Fagioli (1986), Lachiewictz et al. (1985), and Young et al. (1985), include some of the features, but they do not include the neurodegenerative findings. Here, the coexistence of these multiple unusual features and brain abnormalities in siblings suggests a syndrome, more extensive than previously described, with a slowly progressive course.

Author information
Affiliations
Department of Pediatrics, Department of Pediatrics, Medical Student, Southern Illinois University School of Medicine, Springfield, IL, USA, Southen Illinois University School of Medicine, Springfield, IL, USA, Southern Illinois University School of Medicine, Springfield, IL, USA

Roy E Jonas, Virginia E Kimonis & Augusto Morales

Additional information
(Spon by: Donald N. Medearis, Jr., M.D.)

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