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Cerebellomedullary compression in recessive craniometaphyseal dysplasia

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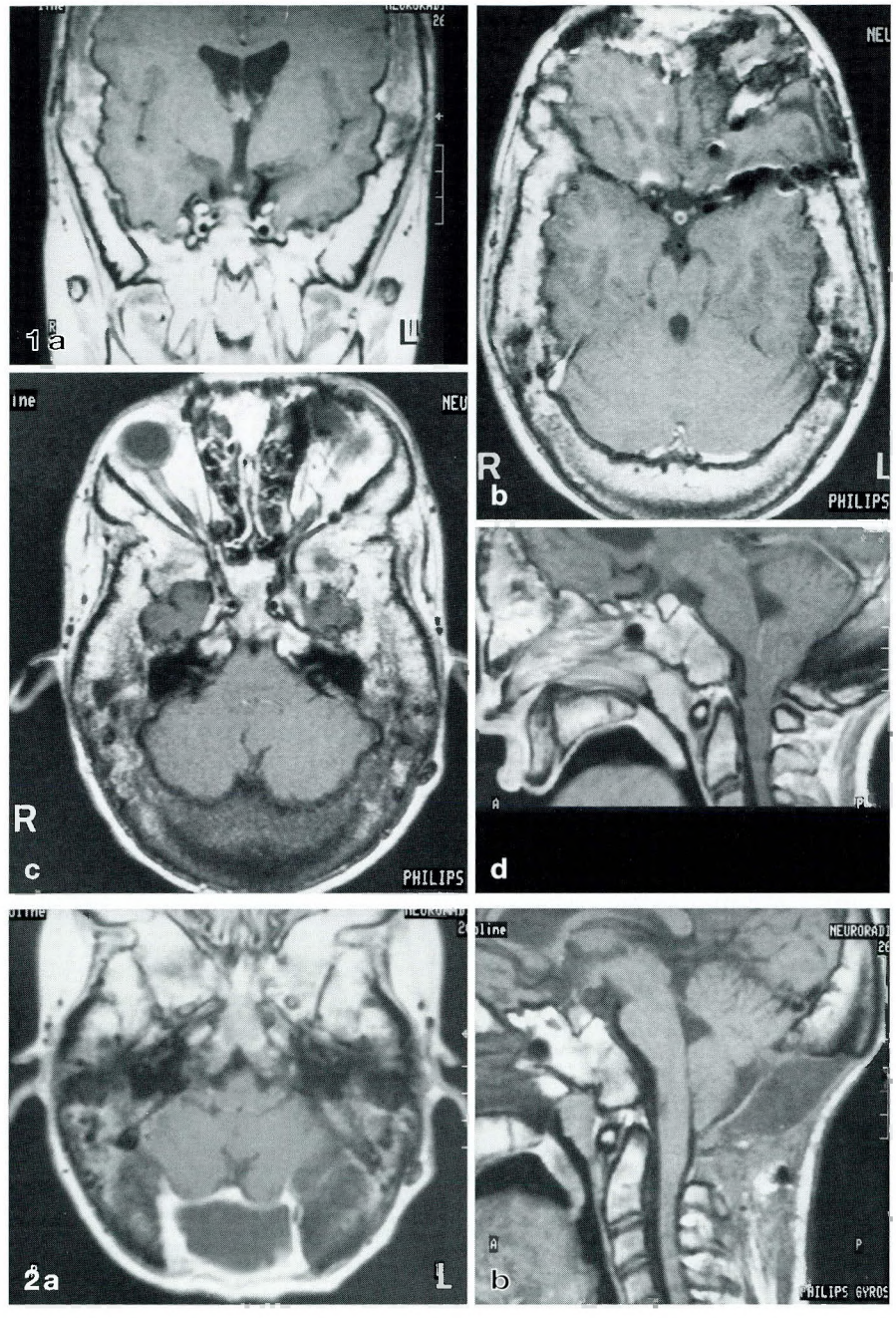


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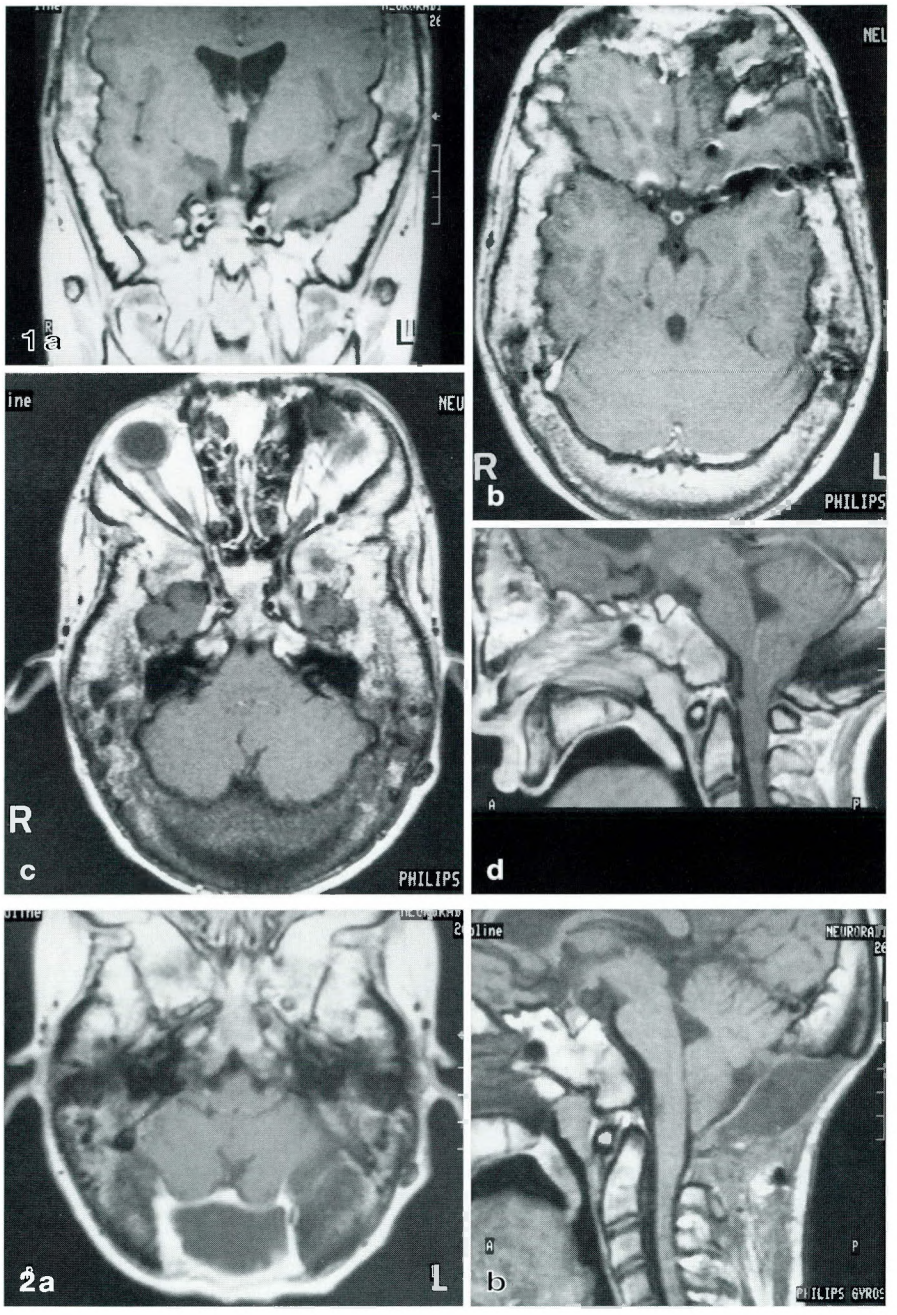


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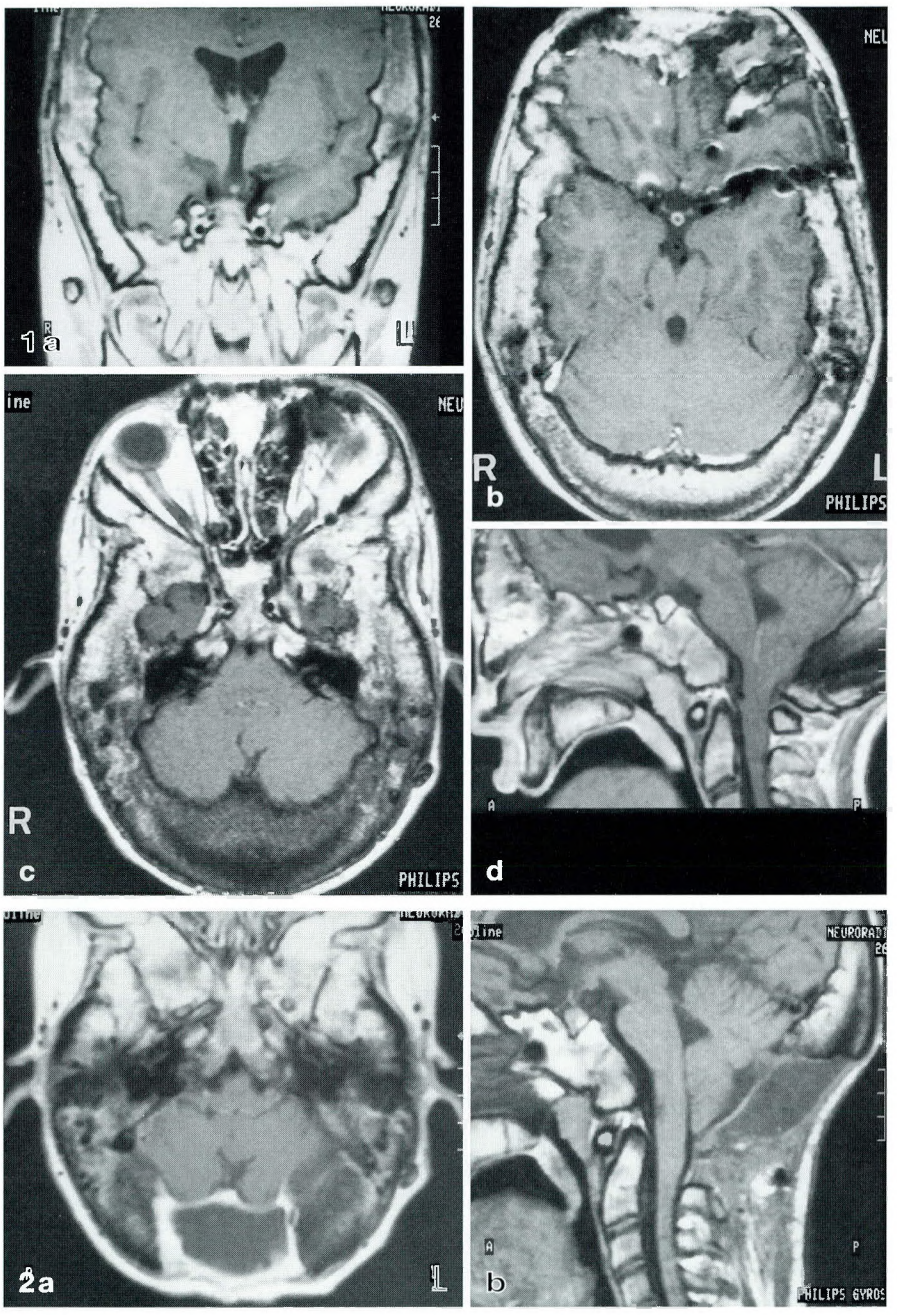


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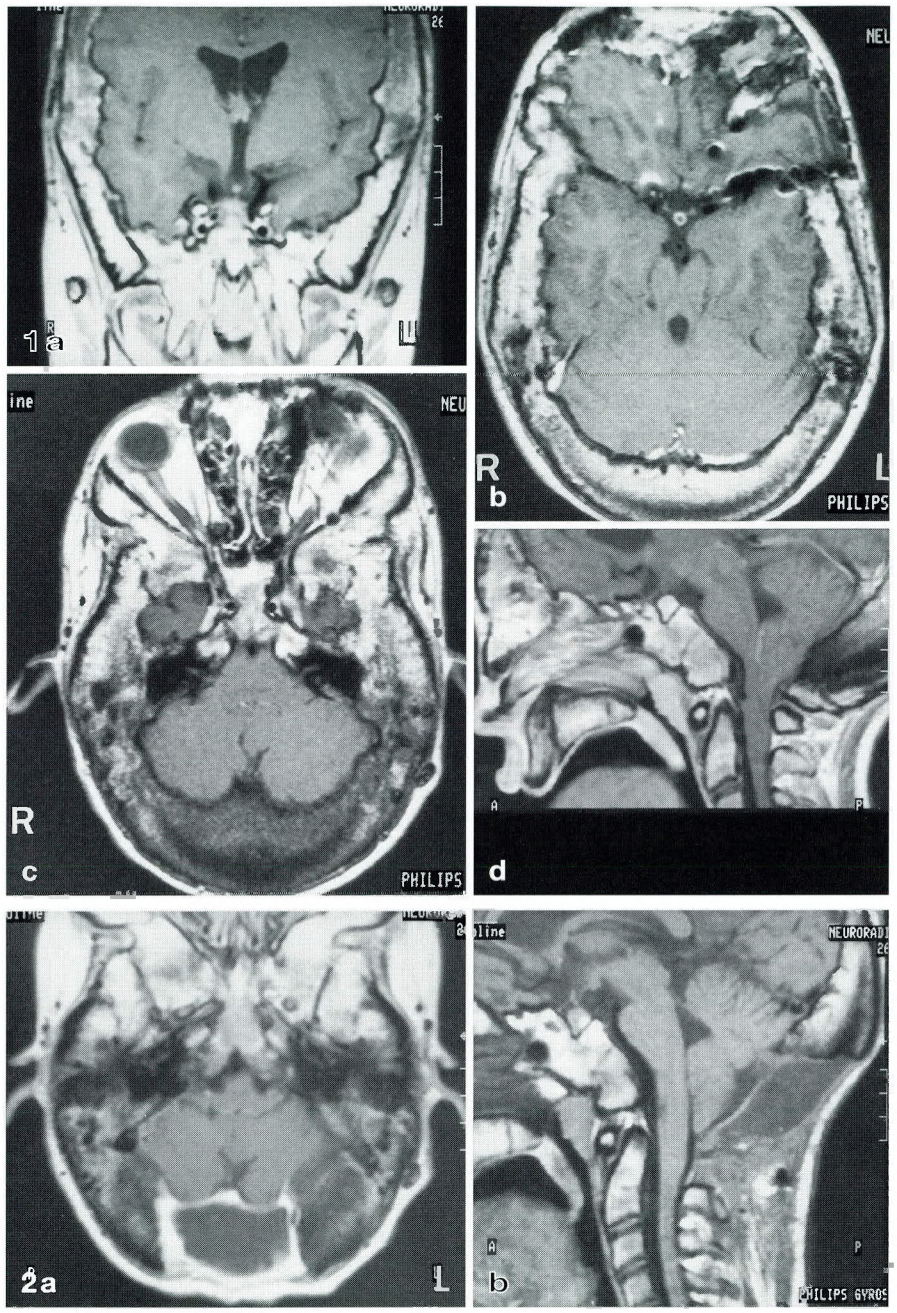


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No medical treatment is available for reducing bony overgrowth in CMD or other similar cranial and tubular bone disorders. In our patient permanent visual impairment has possibly been avoided by optic nerve decompression. Surgical decompression of the facial canal and middle ear were considered technically impossible.

This case illustrates that in severe CMD posterior cranial fossa compression may be a serious problem. Narrowing of the foramen magnum has been reported in other cranial and tubular skeletal dysplasias, particularly progressive diaphyseal dysplasia (Camurati-Engelmann disease) and in sclerosteosis [7, 8]. While visual impairment is unusual in the latter, it typically causes medullary compression and may be complicated by raised intracranial pressure and sudden death.

Our patient has greatly improved following a posterior cranial fossa craniectomy combined with a C1 laminectomy. It is likely that new bone will be formed and it may be that repeat decompression will have to be considered.

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