



A variant case of Meckel Gruber syndrome in a couple with a history of Sandhoff disease

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A 31 years patient G3 P2 with a history of Sandhoff disease in her first baby, with a TOP in a 2nd pregnancy following CVS confirmation of the same disease, had a normal result in the current pregnancy with a normal follow-up. The couple report a 2nd degree consanguinity . She presented at 22 w for a morphology scan with a fetus with the following abnormalities:

- Occipital encephalocele with a wide opening of 15 mm and protrusion of one occipital lobe, alteration of the cerebellum and elongation of the midbrain.
- Enlarged bilateral kidneys with a normal pyelic size and without any cystic image .
- Post axial polydactyly seen in one hand
- Genital ambiguity including female genitalia and enlarged clitoris resembling a penis

No cardiac , facial, or other visceral abnormalities were detected.

This case is interesting because it lacks the classical images in the kidney and because of the different genetic etiology of Sandhoff's disease (chr 5) and Meckel's disease spectrum (chromosomes 8 ; 11 12; 17).

