A RARE DISORDER OF PEROXISOMAL METABOLISM

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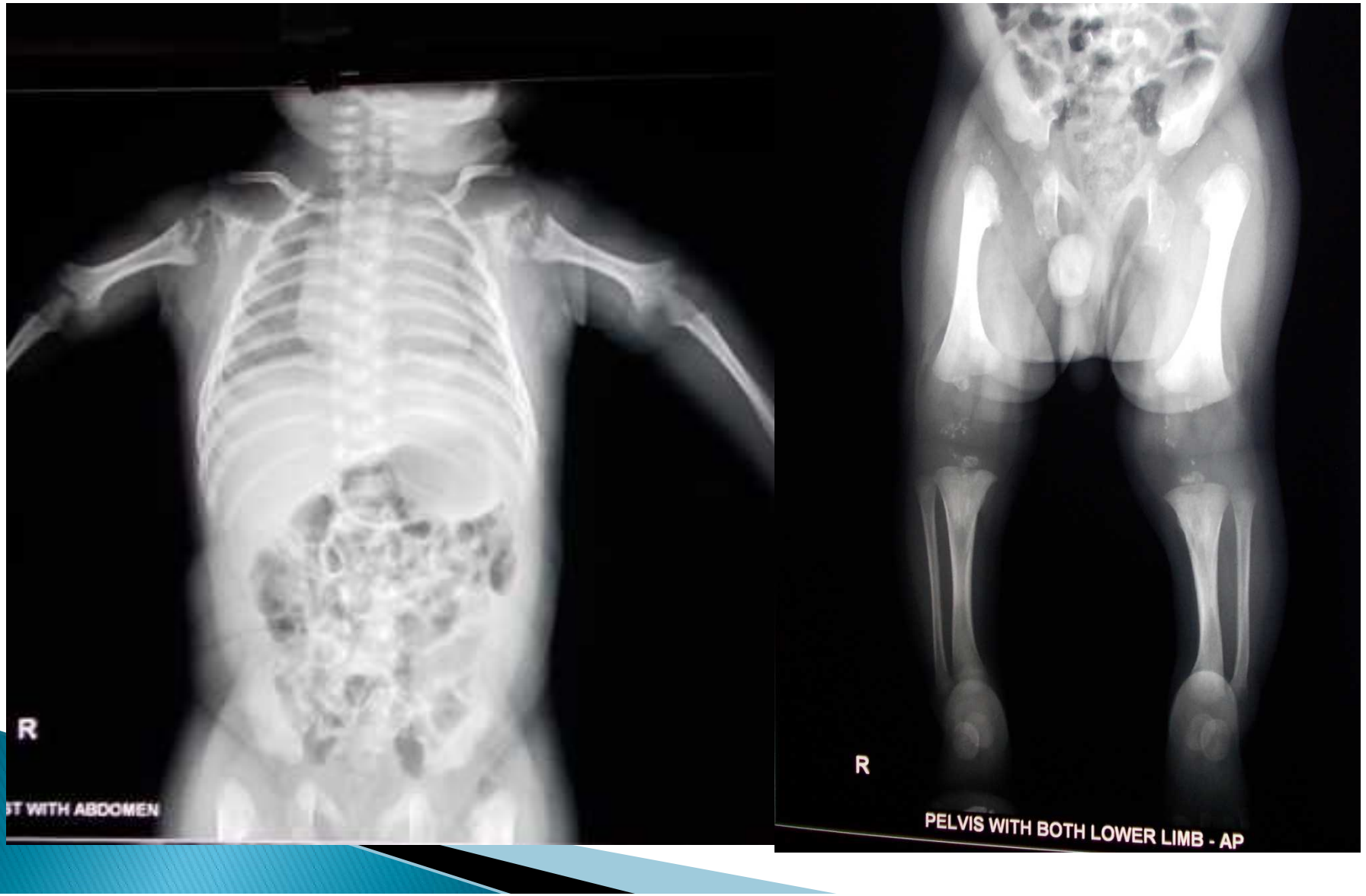
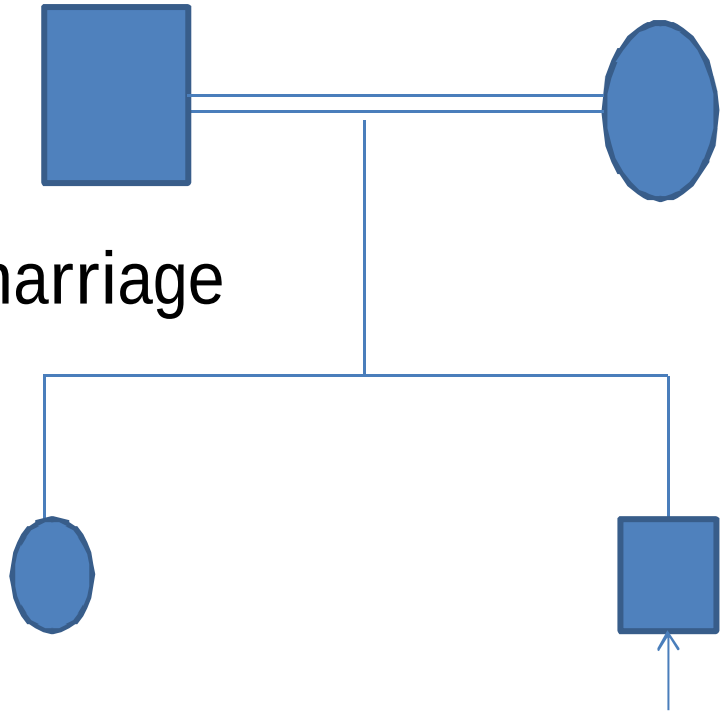
- 1 month old male infant with limb anomalies
- Examination- dysmorphic facies,contractures and rhizomelia.
- Ophthalmological evaluation - bilateral cataract.

X rays showed shortening of humerus,stippling calcification at epiphysis of humerus and femur and metaphyseal cupping.



- 3 rd degree consanguinous marriage



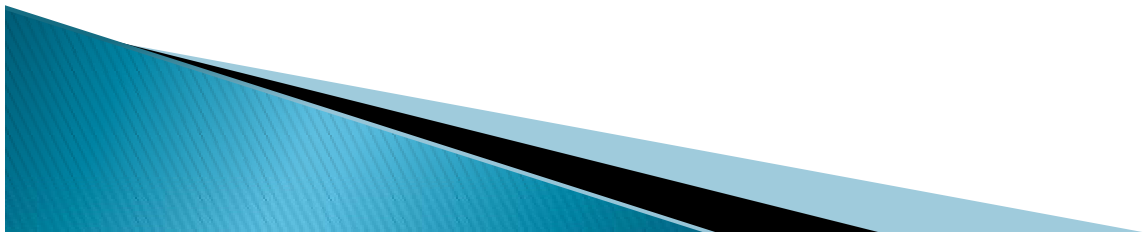
- History , Clinical features and x rays S/O chondrodysplasia punctata

•Biochemistry- Reduced RBC Plasmalogen levels

Normal plasma VLCFA levels

•Mutation analysis - Next generation sequencing showed PEX 7 Intronic variation - homozygous mutation in the baby and heterozygous mutation in the mother.

•Mutations in PEX 7 prevents peroxisomal biogenesis factor 7 from transporting critical enzymes into peroxisomes .



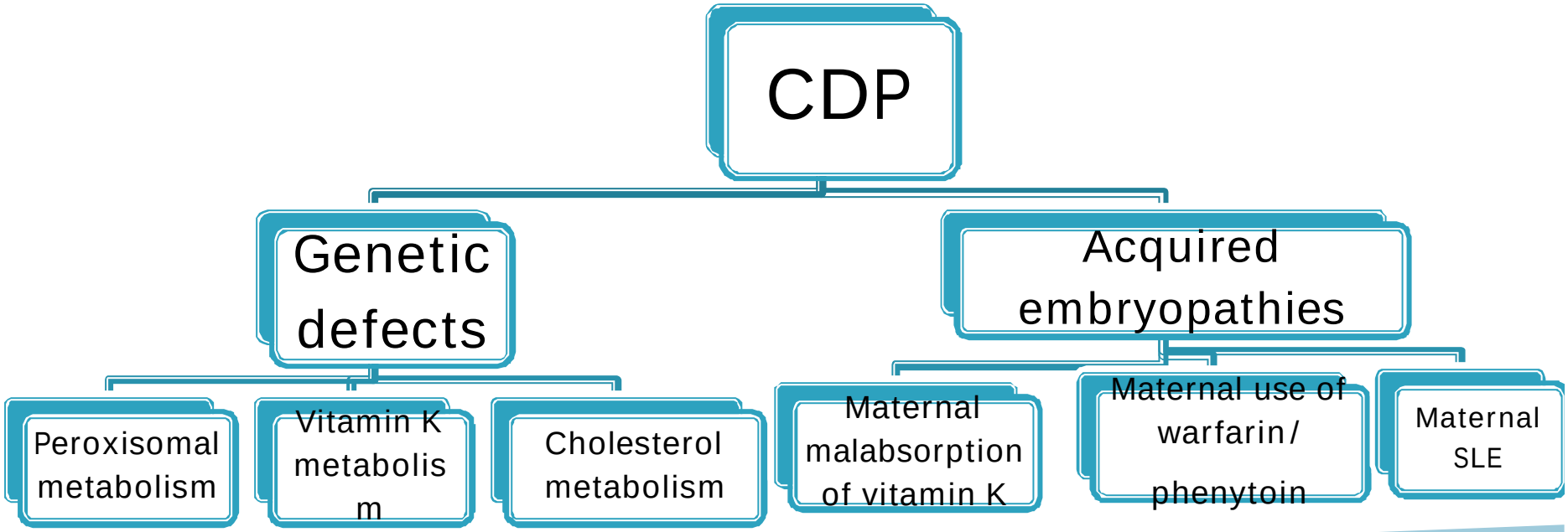
Rhizomelic chondrodysplasia punctata

- Rare peroxisomal biogenesis disorder
- Autosomal recessive inheritance
- Three types- RCDP 1, 2 and 3 – Type 1 most common

- Phenotype-

* Typical facies
* Symmetric proximal limb shortening
* Cataract
* Epiphyseal stippling

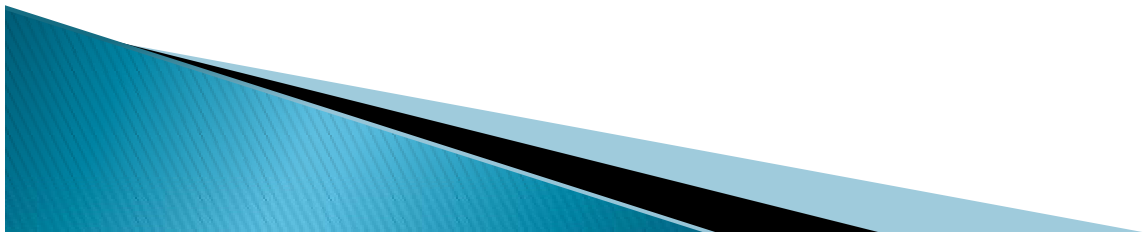
CHONDRODYSPLASIA PUNCTATA



Conclusion

DNA testing for rare disorders is not readily available

However if clinical features and X-rays are suggestive- Biochemical testing would be helpful in clinching the diagnosis.



THANK YOU