

Enfermedad de Lesch-Nyhan, (1984 – 2015)

Algunas consecuencias de la extracción dentaria

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A Familial Disorder of Uric Acid Metabolism and Central Nervous System Function*

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Baltimore, Maryland *Miami, Florida*

Lesch M, Nyhan WL. Am J Med 1964;36:561-70.

Hyperuricemia and Brain Disorder in Children—*Lesch, Nyhan*

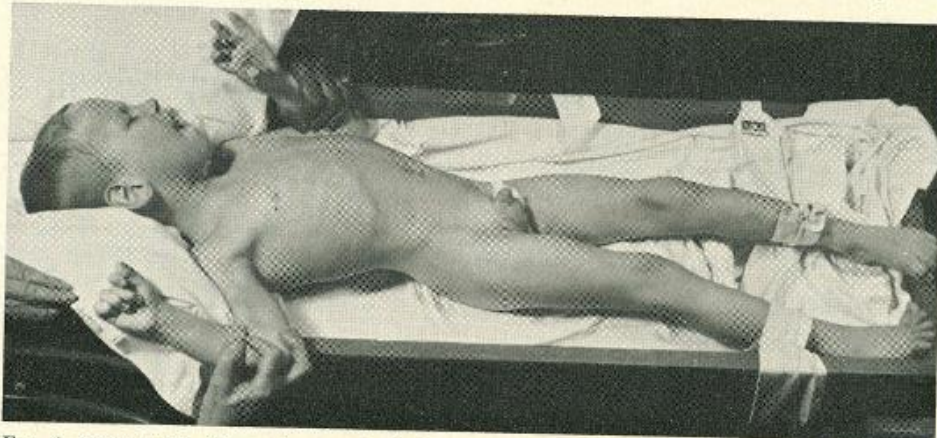


FIG. 1. Patient M. W. at four and a half years of age. The legs are in the characteristic position.

Hyperuricemia and Brain Disorder in Children—*Lesch, Nyhan*

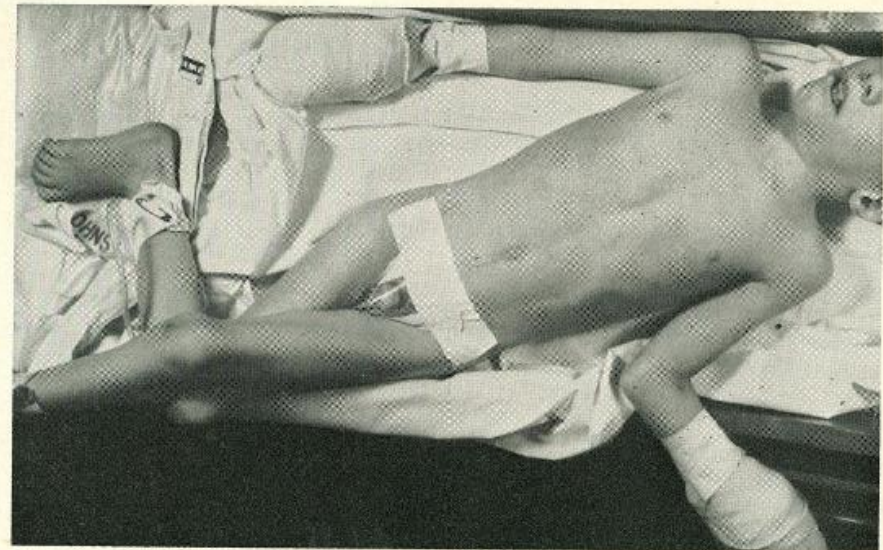
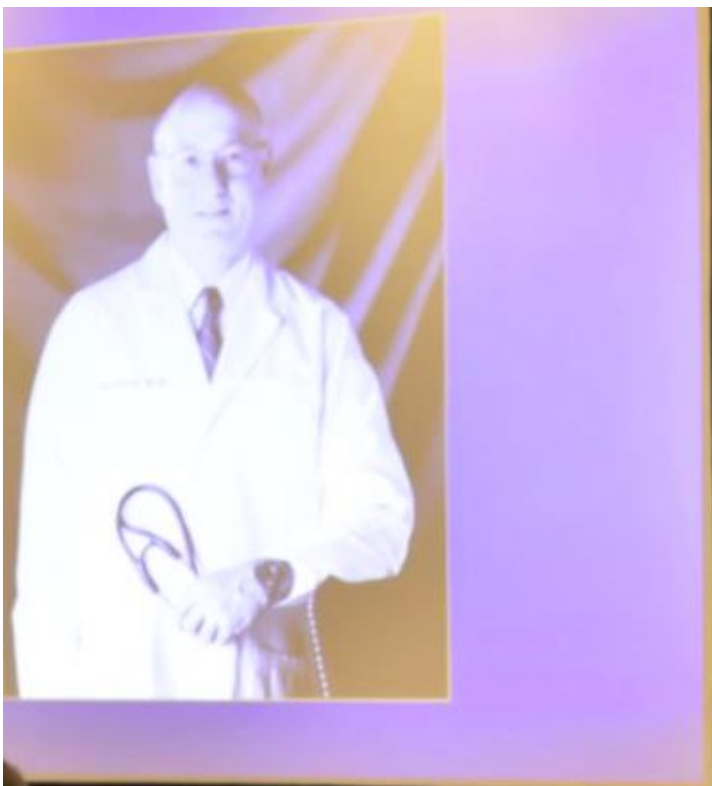
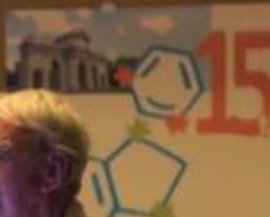


FIG. 2. Patient E. W. at the age of eight years. The mittens on the hands are to protect the child from biting his fingers. The lower lip has been chewed away and is no longer accessible to the central incisors. The marked hypertonia is suggested by the position of the legs.



 **15th** International Symposium
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Enzyme Defect Associated with a Sex-Linked Human Neurological Disorder and Excessive Purine Synthesis

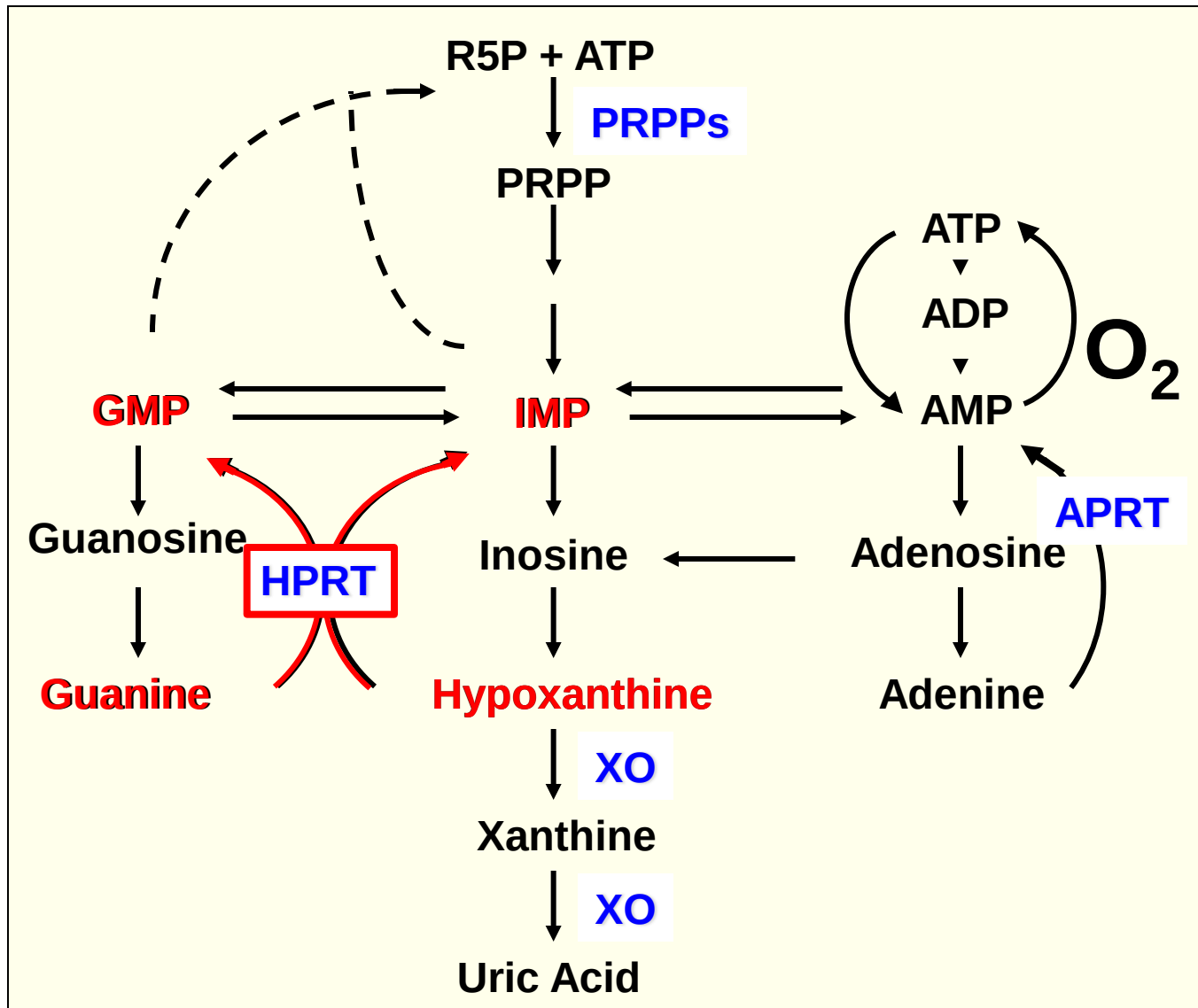
Abstract. A sex-linked familial neurological disease consisting of cerebral palsy, mental retardation, choreoathetosis, and compulsive aggressive behavior is associated with a loss of an enzyme that participates in purine metabolism, namely, hypoxanthine-guanine phosphoribosyltransferase. The production of excessive uric acid in this disorder implies that the enzyme is involved in the normal regulation of purine biosynthesis. This is the first example of a relation between a specific enzyme defect and abnormal compulsive behavior. It is also the first enzyme defect in purine metabolism demonstrated in a neurological disease.

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[Seegmiller JE, et al. Science 1967;155:1682-5.](#)

Purine Metabolism



¿Extracción dentaria completa?

Consequences of Delayed Dental Extraction in Lesch-Nyhan Disease

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Abstract: Patients with Lesch-Nyhan disease (LND) often engage in self-injurious biting. This problem requires difficult management choices, sometimes including removal of the teeth. Although many health care professionals are reluctant to remove teeth in a child because of the permanent negative cosmetic consequences of the edentulous state, disfigurement of the face and tongue from self-biting can be worse. We analyzed the records of 5 LND patients who used mouth guards to spare the teeth. Success was variable, and dental extraction ultimately was required in 4 cases. We also reviewed previously published cases on the use of dental devices to spare teeth in LND. Various devices have been recommended, but failure rates are high, and tooth extraction often is still needed. Although dental extraction is not required in all cases, it should not be delayed when biting is severe.

Dos enfermos tras extracción dentaria completa, al cabo de **12** y **17** meses.

Caso 1: MM.

- 20 m. retraso psicomotriz; orina “*arena de ladrillo*”.
- LND, splicing, exclusión exón 4.

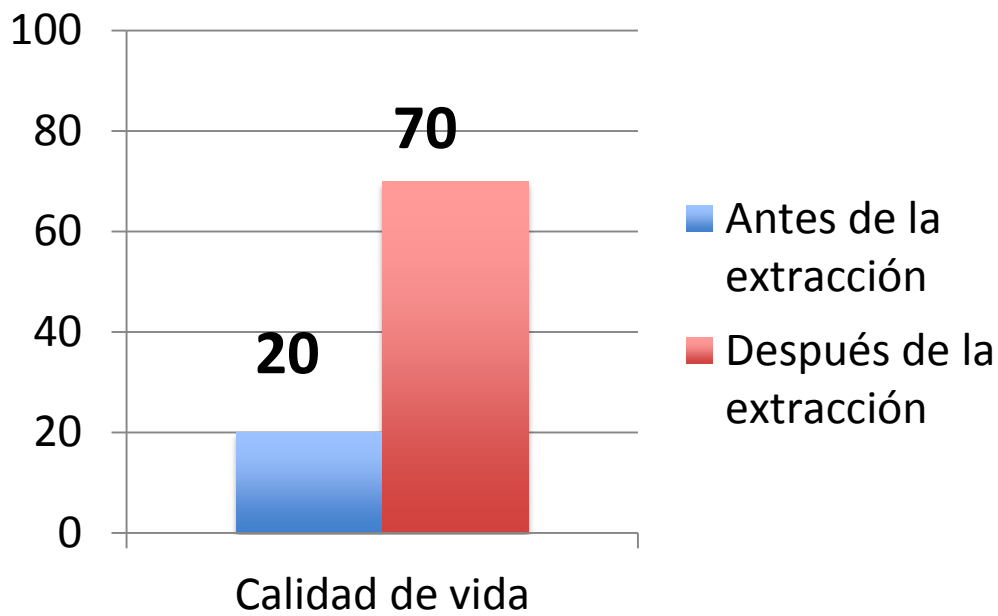


MM - conducta autolesiva

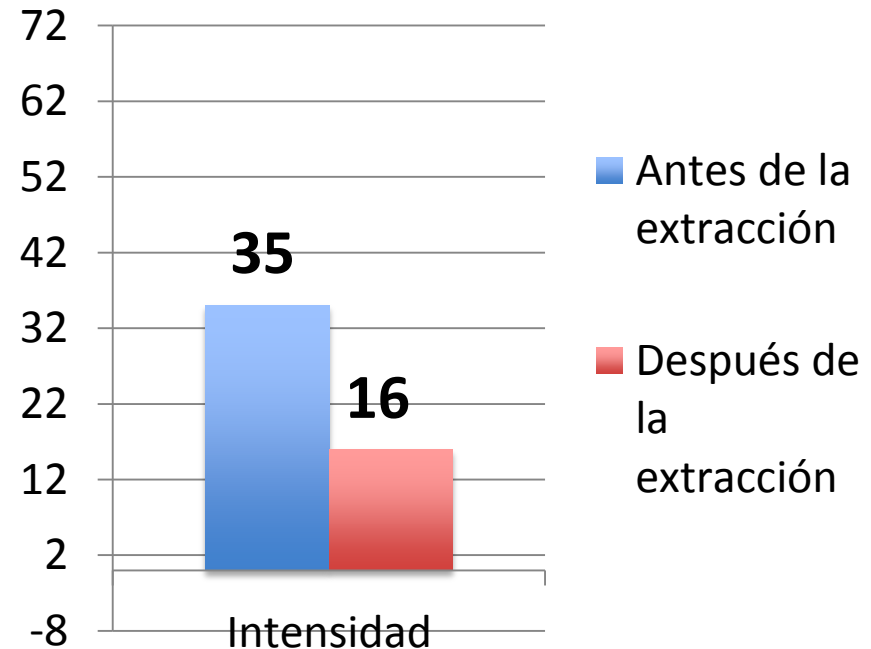
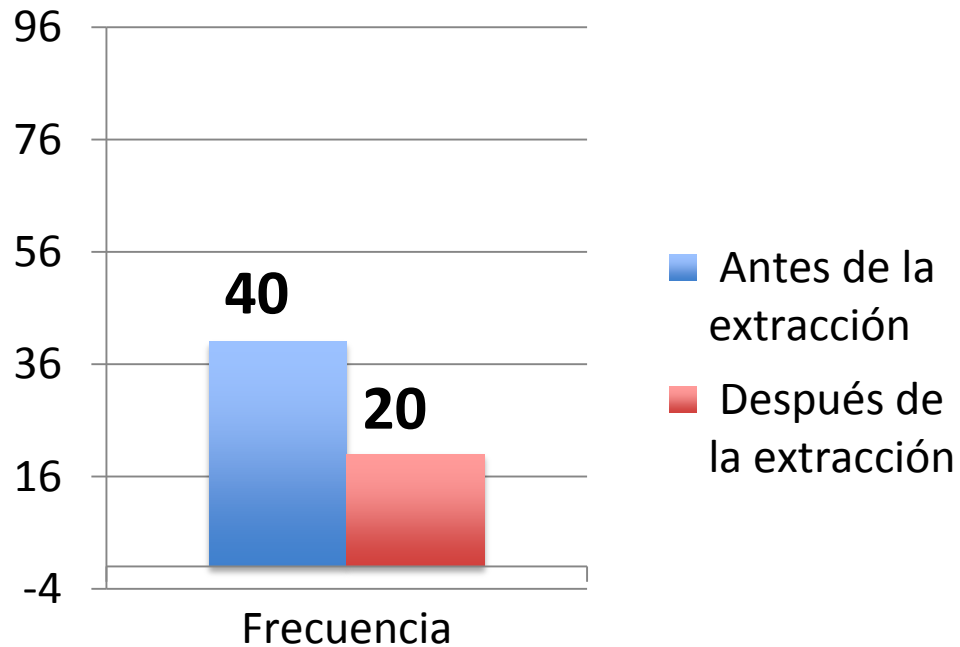


Evolución MM

12 meses



Conducta autolesiva



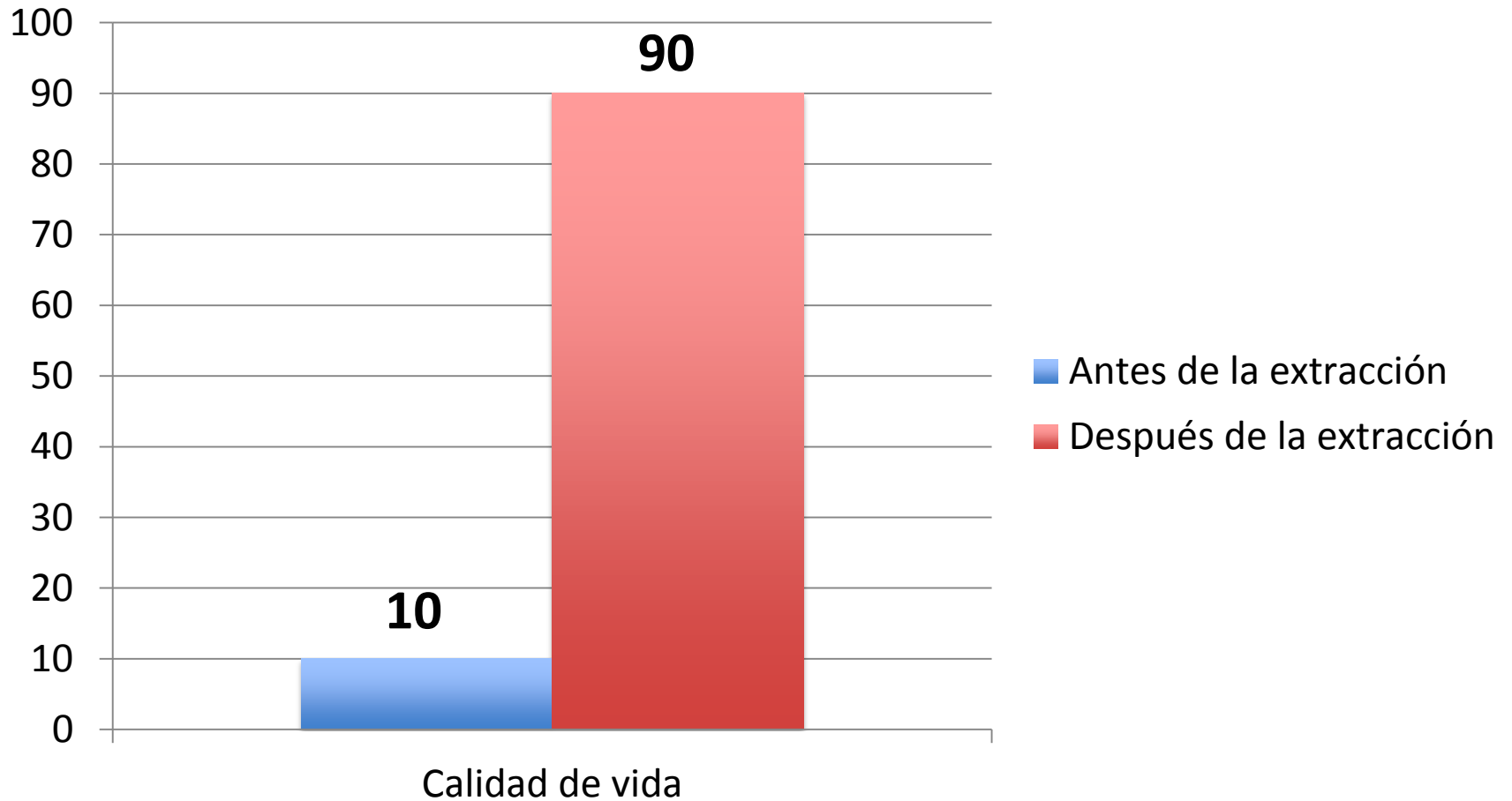


Caso 2: MA

Retraso psicomotriz y eliminación de orina *en polvo de ladrillo*.

10 meses diagnóstico LND: actividad nula de HPRT por delección gen *HPRT1*: exón 1 y región promotora.

Evolución MA



Caso 2: MA evolución **17** meses



Conclusión

La extracción dentaria completa en la enfermedad de Lesch-Nyhan se acompaña de otras formas de autoagresión, **tal vez menos perjudiciales y prevenibles.**



¿ENSEÑANZAS ...?

- **Seguimiento clínico ...**
- **Expandir conocimiento enf. LN.**
- **Medida terapéutica: pros y contras.**

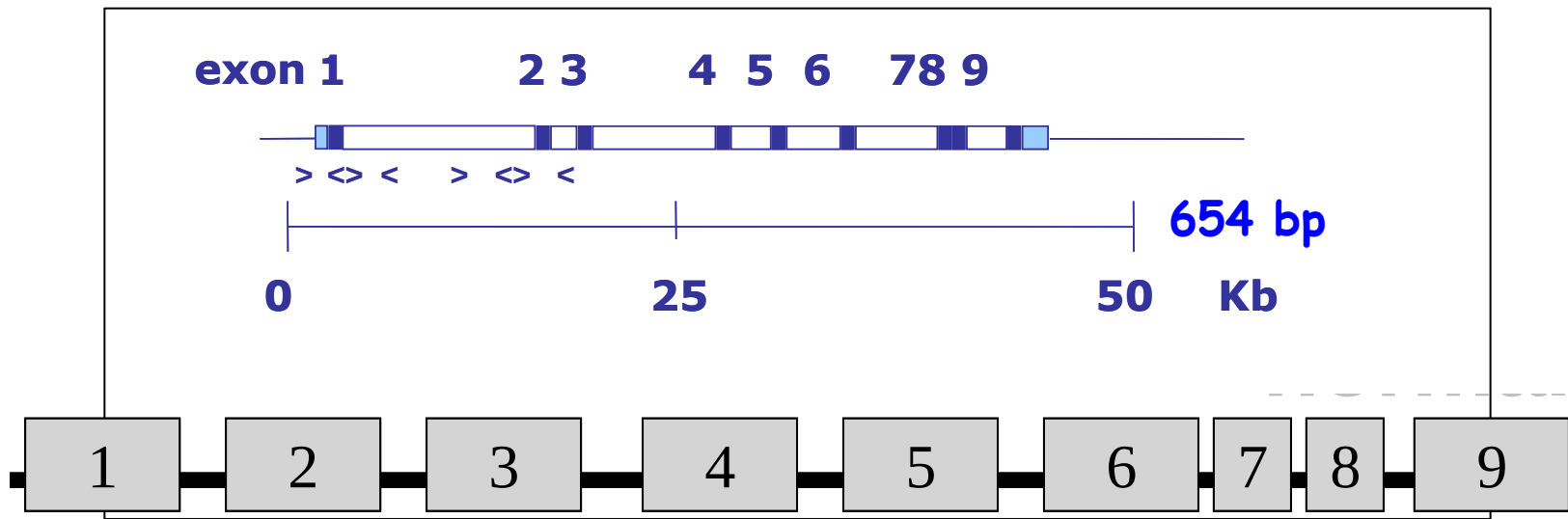
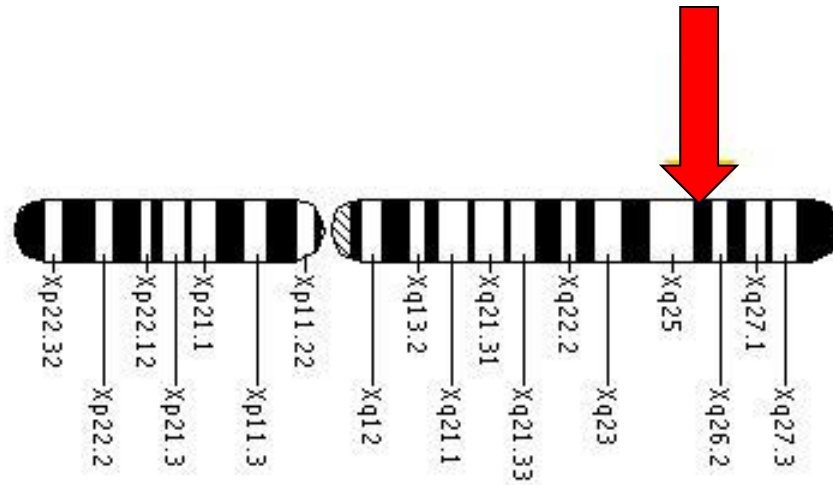
Metabolic – Vascular Unit, Division of Internal Medicina,
La Paz University Hospital, Madrid, **Spain**.



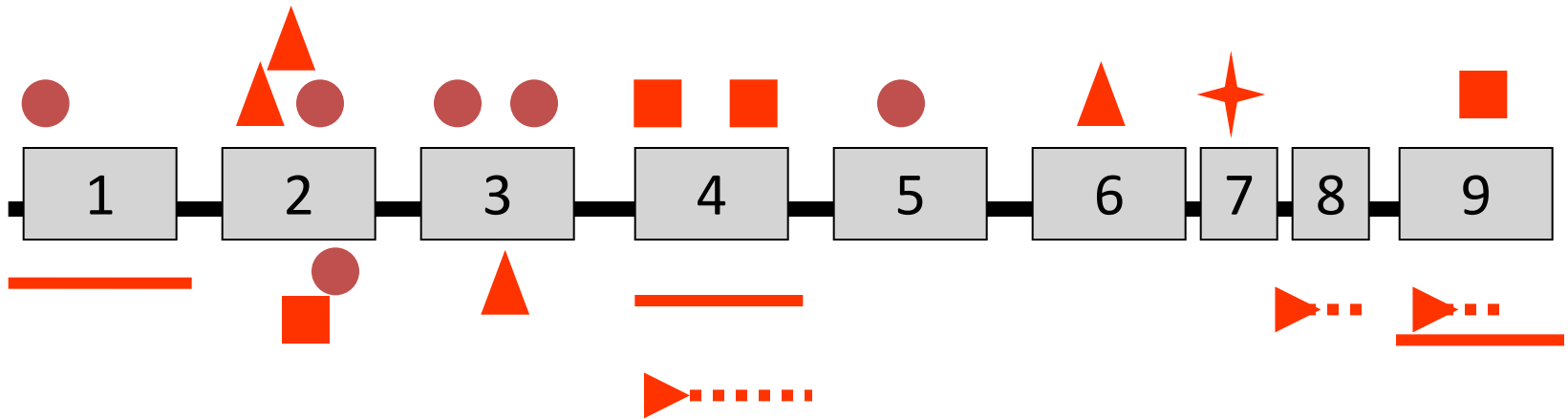
“Building up the future from the past is a very limitanting way to construct the future, because we may assume that whatever could not be in the past will never happen in the future.

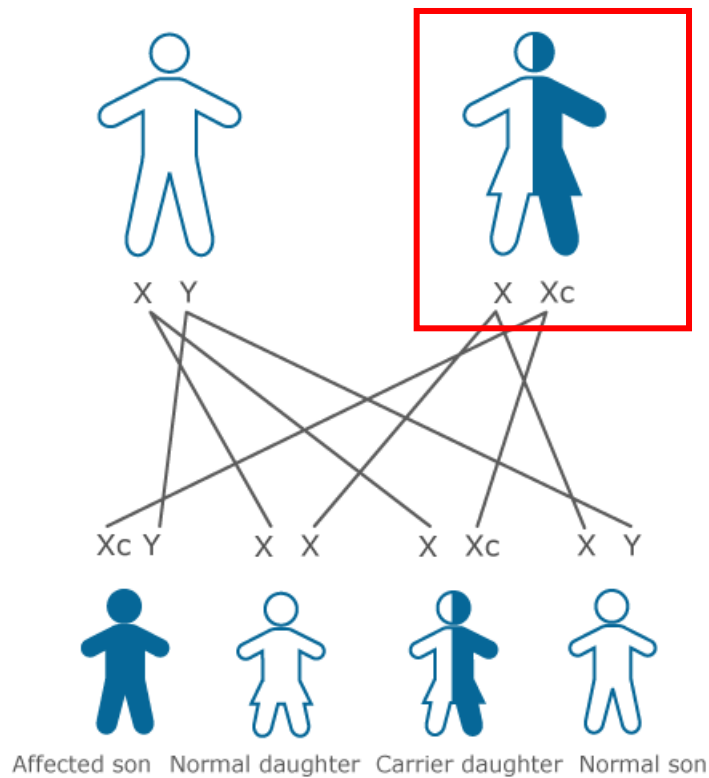
However, building up the future (imagining how the future should be) and then coming back to the present, may be a better way to generate something unpredictable from the past”.

HPRT gene

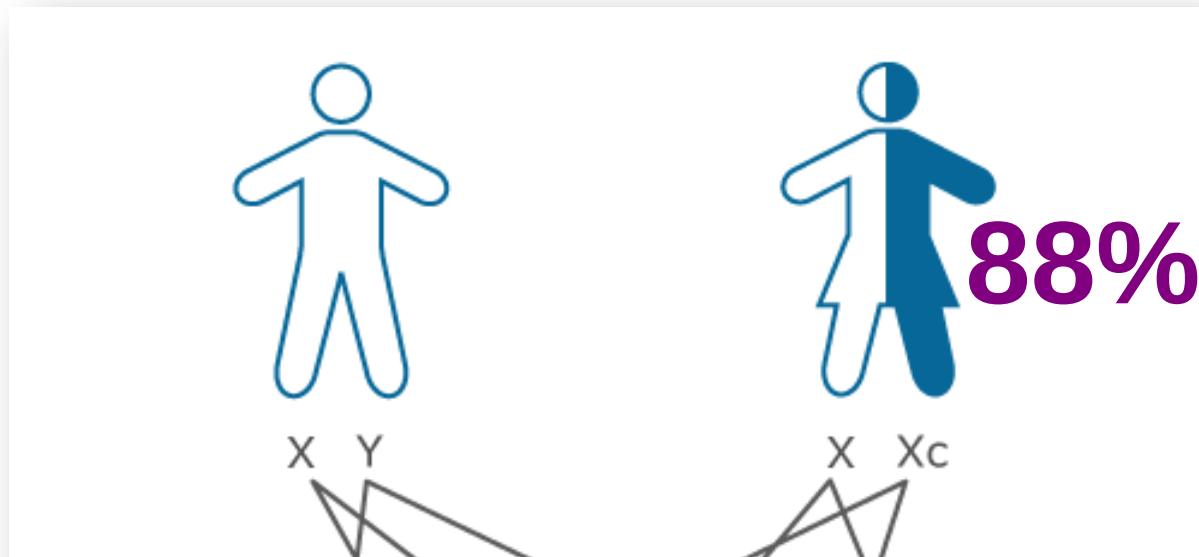


Mutations are different in each family.

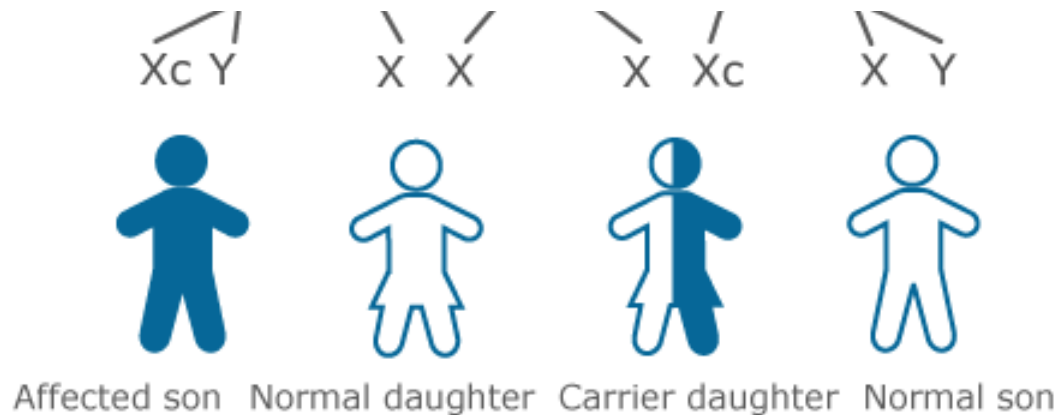




99 females, 33 HPRT def. families



12 prenatal diagnosis: **6** normal males and **3** affected males (25%), and 3 females fetuses



Carrier status

