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MEDICAL GENETICS

It may be said that medical research work in general in South Africa compares favourably with that done anywhere else in the world. This, however, is not the case in the field of medical genetics. We probably lag far behind other countries where most major medical centres have a department of medical genetics.

In the anthropological field we have a number of outstanding genetic workers, and the liaison between genetics and anthropology has been successfully fostered by the Institute for the Study of Man in Africa, founded in 1958 in honour of Prof. Raymond Dart. Other genetic units have been developed by the Institute of Agriculture at the Universities of Bloemfontein, Pretoria, and Stellenbosch, and by the Veterinary Institute at Onderstepoort near Pretoria. Within the province of medical services it may be said that human genetics was introduced to South Africa by Dr. J. G. Davel in 1944. In the field of psychiatric genetics Prof. L. Hurst has long been an advocate for a genetic discipline in psychiatry and medicine, and he has been the moving force behind the recent establishment of a genetic clinic in Johannesburg.

In 1955 Sheldon Reed said: 'Most physicians are called upon to give genetic counselling from time to time. The patient expects the answers he gets regarding heredity in his family to be of the same high quality as the surgical or other medical counselling which he receives. Most physicians, however, have had little training in medical genetics and may not be able to give professional service in this field. The reason that training in medical genetics is still inadequate is that the subject is new, and space in the medical curriculum has not yet been found for it in very many medical schools.'

Much of what Reed said in 1955 applies today, and in fact more so, for advances in biochemistry and in cytogenetics in the last decade have widened the scope of the clinical geneticist and have emphasized the need for genetic clinics within any hospital service; they have also underlined the necessity for training in medical genetics within our curricula.

Apart from the 'utilitarian' aspect — that a counselling service should be made available — it seems to us that there is a need for a clinical genetics unit to deal with medical genetics in the broadest sense.

Such a unit was formed in Johannesburg just over a year ago. At the moment the unit is under the aegis of the Department of Psychiatry, and an office within that department is set aside for the purpose. There is available: (1) A secretary, (2) a filing system for (*a*) patients' notes and family-trees and (*b*) reprints and recent literature, (3) facilities for examination of and discussion with patients, and (4) a library of books on genetics which is being built up.

The aims of this unit may be said to be:

1. To provide a *heredity counselling service* and in this way help at the personal level and in the wider sense to forward the aims of eugenics. The service is quite free and patients are usually referred by a member of the profession who has been presented with a problem of heredity in that particular family. The fact that many of the cases come from outside Johannesburg, outside the Transvaal and even from the Rhodesias points to the need for more clinics of this nature.

South Africa is a fertile field for familial problems as indicated by the work of Dean on porphyria and Klintworth on Huntington's chorea. A population explosion has occurred in this country — a nation has grown from a few original ancestors in some 300 years!

2. To provide a *reference system* for the future:

(*i*) Collection of family trees.

(*ii*) Documentation of all forms of genetic disorder seen in South Africa.

(*iii*) The creation of a genetic reference library, i.e. recent and classic books on the subject.

(*iv*) A filing system of the relevant recent reprints and journals pertaining to the subject.

(*v*) With the above at hand, to provide the nucleus of a teaching centre in medical genetics both through lectures and the publication of articles.

3. To *study and document* all forms of genetic disorder. This programme embraces *research* aspects and encroaches on several other disciplines. Thus, collaboration would be sought with the cytogenetics unit at the South African Institute for Medical Research, with the biochemist, in locating the site of an enzyme block, and with the haematologist-immunologist in carrying out full blood-group studies, etc. As examples of this activity the following can be mentioned:

(*i*) A survey for phenylketonuria is being carried out by Dr. I. Anderson at Witrand Mental Institution.

(*ii*) A recent visit to the 'Hell' was organized to investigate a 'genetic isolate' and all the implications of this for society.

(*iii*) A survey of mental defectives for sex-chromosome abnormalities, etc., is proposed.

Elsewhere in this issue of the *Journal* we publish articles dealing with diverse aspects of medical genetics.

DIE DRINK VAN SEEWATER

Verhale in tydskrifte en koerante van oorlewendes van seerampe beklemtoon dikwels die gevare verbonde aan die drink van seewater. Hierdie ondervinding van sulke oorlewendes word natuurlik gestaaf deur die bekende fisiologiese feite: Die soutinhoud van die liggaamsvloeistowwe is ongeveer 1%. In die oseaan is die soutinhoud van water ongeveer 3·5%. Tydens goeie voeding en optimale gesondheid is die niere in staat om die liggaam se soutkonsentrasie tussen baie enge perke te reguleer. As water egter onthou word, is die gemiddelde sout-

uitskeiding selde hoër as 2%. Die inname van 'n 3·5% oplossing sou dus beteken dat die niere slegs 'n 2% oplossing kan uitskei en geleidelik sal die soutkonsentrasie van die liggaam styg. Hierdie hipertoniese seewater-inname lei tot 'n hipertoniese oplossing in sirkulasie wat water vanuit weefsel onttrek om die onkotiese druk tussen weefsel en vaskulêre kompartement te herstel. Die gevolg is toename in bloedvolume, en 'n diurese volg. Die netto resultaat is 'n dehidrasie van weefsel met daaropvolgende versteuring van waterstofioon-konsentrasie, styging van bloedureum en plasma-proteïnkonsentrasie. Die kardiale omset verminder, dors tree in, en uiteindelik volg uitputting, skok, en die dood.

Dr. Alain Bombard het egter in 1952 die Atlantiese Oseaan oorgesteek in 'n luggevulde vlot, 'n reis wat altesaam meer as twee maande geduur het. Hy het hoofsaaklik seewater gedrink, aangevul deur vloeistof uit visse te pers. Sy mening was dan dat mens seewater in klein hoeveelhede mag inneem om dehidrasie teen te gaan en dat vars water wat beskikbaar is liefs sorgvuldig bewaar moet bly vir later as die toestand kritiek word.

Hierdie bewerings is gestaaf deur dr. G. Aurey, hoof mediese beampte van die Franse Vloot wat in 1953 en 1954 twee- tot vier-daagse eksperimente uitgevoer het op vrywilligers, onder wie dr. Aurey self. Die proefkonyne kon hul werksaamhede onmiddellik hervat.

'n Duitse geneesheer, Lindemann, 'n veteraan Atlantiese vaarder, wat die oseaan drie maal in kano en rubberboot trotseer het, het egter die bevindings van Bombard en Aurey weerspreek. Hy stel dit dogmaties dat seewater *nooit* gedrink moet word nie, en vir die uitdruk van visse het 'n mens 'n meganiese pers nodig om doeltreffend te wees. Geen vis moet geëet word as varswater nie beskikbaar is nie.

In 1959, vanweë wye publisiteit aan die eksperimente van Bombard en Aurey, was daar baie verwarring. Fisiologies klink dit na kettery, maar per slot van rekening is dit tog ervaar. Reëls vir skipbreukelinge kan egter nie deur die betrokke owerhede neergelê word op 'n weerspreekte ervaring nie, en soos gebruiklik is die probleem na 'n langnamige komitee verwys. Die Inter-regerings Maritieme Konsultatiewe Organisasie het dit weer verwys na die Wêreld Gesondheidsorganisasie, wat 'n komitee benoem het bestaande uit deskundiges van Switserland, Frankryk, die Verenigde Koninkryk, en die V.S.A. Hulle verslag[2] word saaklik opgesom in 'n onlangse bespreking.[1]

Die uitwerking van die drink van seewater kan in drie kategorieë verdeel word:

(1) *Die uitwerking op die derm.* Die katarse wat volg op inname van soute van magnesium en natrium is welbekend en, na gelang van die dosis ingeneem, mag seewater dus lei tot krampe en uitgesproke diaree. Die uitwerking wissel tussen individue, maar is meer geneig om na groot hoeveelhede voor te kom. Dit sal natuurlik lei tot 'n verergering van dors as gevolg van waterverlies.

(2) *Die uitwerking op die liggaam as geheel.* Die oorlaaiing van die sirkulasie met sout kan alleen bekamp word deur 'n osmotiese diurese wat dehidrasie van weefsels vererger. As dit nie doeltreffend bekamp word nie, neem bloedvolume toe met ewe nadelige gevolge.

(3) *Die uitwerking op die geestestoestand.* Daar is, volgens hul bevinding, afdoende bewys dat selfs geringe hoeveelhede seewater sommige individue nadelig mag beïnvloed, en groter hoeveelhede lei dikwels tot geestesversteuring en selfmoord-neigings.

Die vraag of varswater met klein hoeveelhede seewater gemeng kan word om die voorraad langer te laat hou, is teoreties moontlik in mense en is eksperimenteel in diere aangetoon, maar die komitee vind geen aanvaarbare bewyse om hierdie 'n praktiese voorstel te maak nie.

Die argument wat aangevoer word dat die drink van seewater die moraal kan onderskraag, 'n belangrike oorweging in die middel van die oseaan, is ook nie oortuigend vir die komitee nie. Hulle meen dat meer bereik kan word deur aan slagoffers te verduidelik dat die dood as gevolg van 'n tekort aan water beswaarlik sal intree voor etlike dae. Eksperimente toon dat gesonde mense ses dae lank redelik fiks kan bly sonder water, en mense het al vir dubbel dié tydperk die gevolg van skipbreuk sonder water oorleef. Verder is aangetoon dat 500 ml. water (1 pint), aangevul deur 100 G. koolhidraat, 'n mens ses dae lank sonder enige aantoonbare agteruitgang kan onderhou.

Hulle advies vir skipbreukelinge is dus: ‚Moet *nooit* seewater drink nie. Moet *nooit* seewater met varswater meng nie, veral as dit skaars is. Seewater is gebruik om die mond klam te hou, maar die versoeking om dit in te sluk mag onweerstaanbaar word en die gebruik word dus afgeraai. Moet nooit uriene drink nie.'

Die ondersoek bevestig dus die aanvaarde fisiologiese beginsel dat 'n *water*-tekort alleen deur water en nie deur soutoplossing herstel kan word nie. Die gevolge is dan ook die klinies-bekende gevolge van oormatige hipertoniese vloeistof-toediening. Die waarheid is dus getoets en bevestig.

1. Redaksionele kommentaar (1962): Nature, **196**, 1045
2. Verskeie skrywers (1962): W.H.O. Chronicle (September), **16**, 9.